

# Congenital Heart Disease

## An Approach for Simple and Complex Anomalies



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## Disclosures

- None

## ASCeXAM

- Contains questions on general congenital heart disease, not “adult” CHD
- Study guide contain all of the information in this talk plus addition topics that will be helpful for the exam
- There have been a few questions on fetal echo which have appeared on the ASCeXAM which are covered in the handout
- Insider information provided in study guide – topics that have appeared on prior exams (last page of study guide)

## Which heart defect is more common?

- ★ 1. Tetralogy of Fallot
- 2. Transposition of the Great Arteries
- 3. Aortic Stenosis
- 4. Coarctation of the Aorta
- 5. Atrioventricular Canal

## What is the most common defect seen with Down syndrome (trisomy 21) ?

1. Patent Ductus Arteriosus
2. Tetralogy of Fallot
3. Aortic Stenosis
- ★4. Ventricular septal defect
5. Coarctation of the Aorta

## Congenital Heart Disease

Spectrum of Congenital Heart Disease - Incidence

- 0.5–0.8% of live births \*
- >30,000 individuals/year in U.S.
- 50% simple shunts (ASD, VSD, PDA)  
20% simple obstruction  
30% complex

\* Excludes MVP (4-6%) & Bicuspid AV (1-2%)

## Congenital Heart Disease

Spectrum of Congenital Heart Disease - Frequency

<u>Cardiac Malformation</u>	<u>% of CHD</u>	<u>M:F Ratio</u>
Ventr. Septal Defect	18-28	1:1
Patent Ductus Arter.	10-18	1:2-3
Tetralogy of Fallot	10-13	1:1
Atrial Septal Defect	7-8	1:2-4
Pulmonary Stenosis	7-8	1:1
Transp. of Grt. Art.	4-8	2-4:1
Coarctation of Aorta	5-7	2-5:1
AV Septal Defect	2-7	1:1
Aortic Stenosis	2-5	4:1
Truncus Arteriosus	1-2	1:1
Tricuspid Atresia	1-2	1:1
Tot. Anom Pulm Veins	1-2	1:1

## Congenital Heart Disease

Common Syndromes/Chromosomal Anomalies

<u>Anomaly</u>	<u>Associations</u>
Trisomy 21	VSD, AV Canal
Trisomy 18	VSD, PDA
Trisomy 13	VSD, PDA, Dextrocardia
Turner	Coarctation, AS
Noonan	PS, HCM
Williams	Supravalvar AS, Peripheral PS
Holt-Oram	ASD
Marfan	Aortic root dilation, MVP
DiGeorge	VSD, arch anomalies, TOF



## Congenital Heart Disease

- Currently over 1 million patients over 18 with CHD are alive in this country
- This increases at 4-5% per year
- 20,000 cardiac surgical procedures for CHD/year
- >90% of children with CHD survive to adulthood
- The majority of adult CHD patients will be post-op
- The ASCeXAM does not cover much post-op disease, but has asked questions about operations

## Congenital Heart Disease

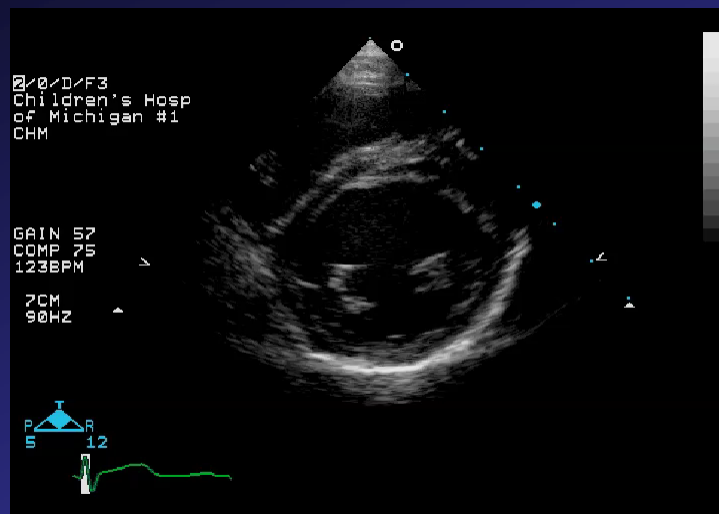
### A Brief History of Operations

<u>Year</u>	<u>Physician</u>	<u>Procedure</u>
1938	Gross	Ligation of PDA
1944	<u>Blalock, Taussig</u>	Syst.-pulm. shunt
1945	Gross, Crafoot	Repair of coarctation
1946	<u>Potts</u>	Aorta to LPA shunt/direct anastomosis
1952	Muller	Pulm. artery band
1953	Gibbon	Repair of ASD
1954	Lillehei	Repair of VSD
1954	<u>Glenn</u>	SVC-PA shunt
1956-7	<u>Mustard/Senning</u>	Atrial correction of TGA
1955	Lillehei, Kirklin	Repair of tetralogy of Fallot
1960	<u>Waterston</u>	Aorta to RPA shunt/direct anastomosis
1964	<u>Rastelli</u>	Conduit replacement of PA
1967	<u>Rashkind</u>	Balloon atrial septostomy
1971	<u>Fontan</u> , Kreutzer	Repair of tricuspid atresia
1976	<u>Jatene</u>	Arterial switch for TGA
1978		Cold blood cardioplegia

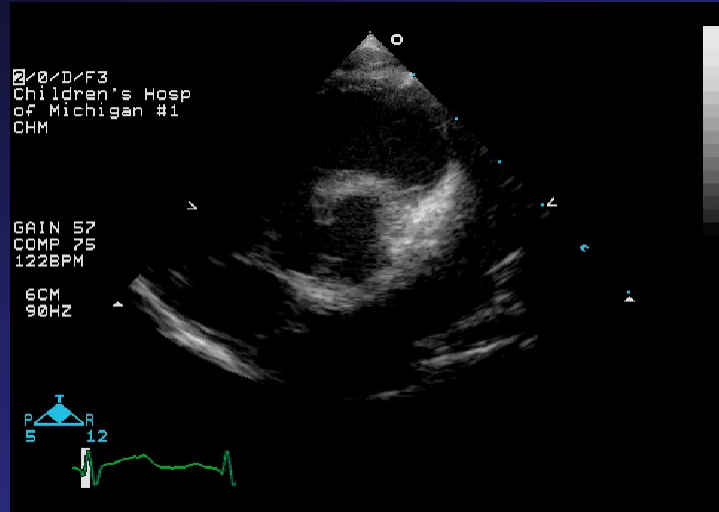
## Case 1 - 3 month old with a murmur, tachypnea and failure to thrive



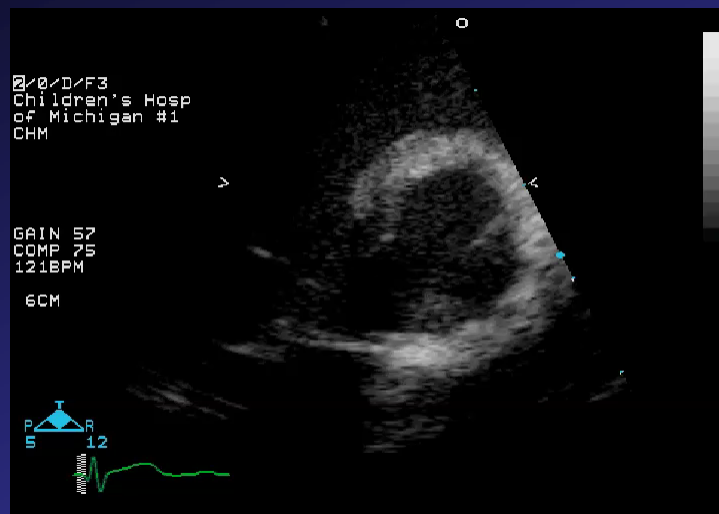
## Case 1 - 3 month old with a murmur, tachypnea and failure to thrive



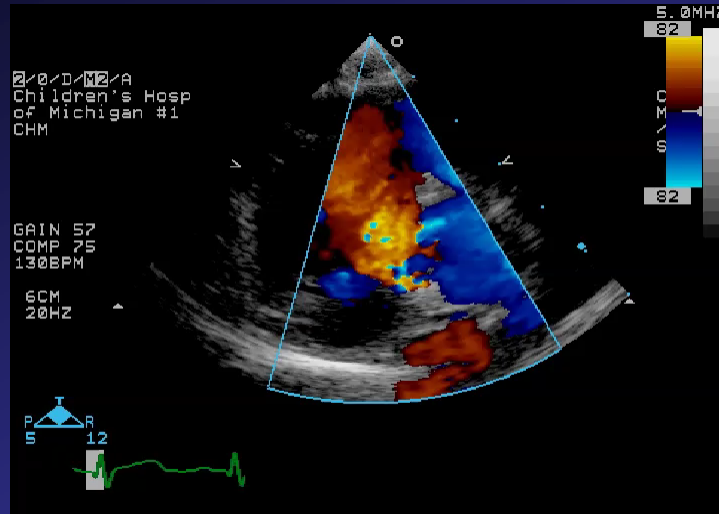
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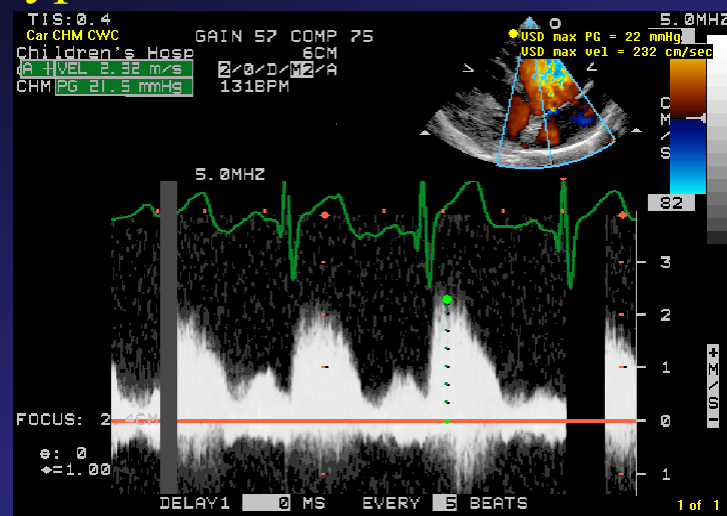
## Case 1 - 3 month old with a murmur, tachypnea and failure to thrive



## Case 1 - 3 month old with a murmur, tachypnea and failure to thrive



## Case 1 - 3 month old with a murmur, tachypnea and failure to thrive



The defect shown in this example is:

1. Secundum VSD
2. Sinus Venosus VSD
- ★ 3. Perimembranous VSD
4. Inlet VSD
5. Supracristal VSD

An isolated VSD will generally produce enlargement of which chamber(s):

- ★ 1. Left atrium, Left ventricle
2. Right ventricle
3. Right ventricle, pulmonary artery
4. Aorta
5. Right ventricle, right atrium

## What is the right ventricular pressure?

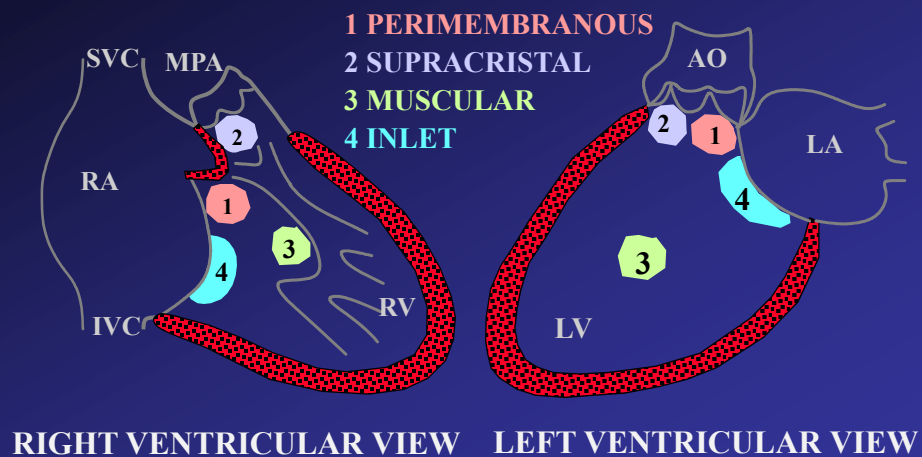
1. Normal
2. Supra systemic
- ★ 3. Systemic
4. Can't tell from information given
5. Want to go home now

## Ventricular Septal Defect

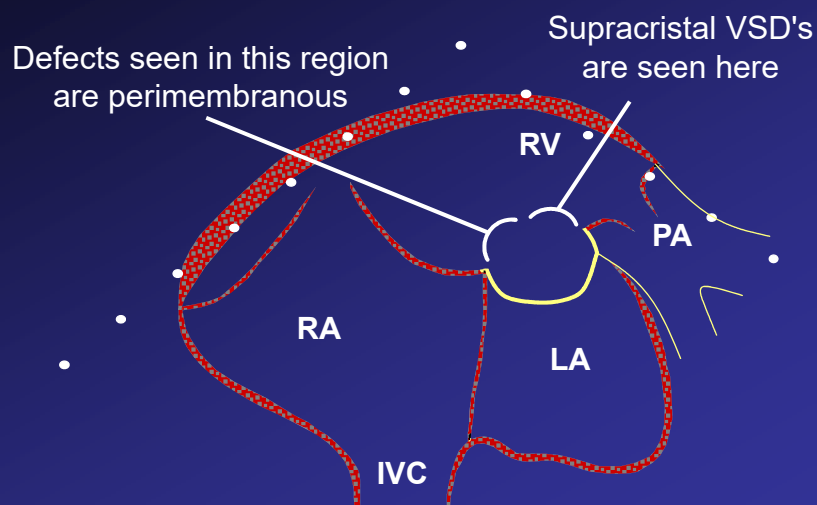
### Clinical

- Most common defect, 25% of CHD
- Shunt flow usually left to right in infants and children
- Symptoms depend on the size of the defect
  - Large - >50% of aortic annulus size
  - Medium - 25-50% of annulus size
  - Small - <25% of annulus size
- Large VSDs result in pulmonary edema
  - tachypnea, poor feeding, failure to thrive in infants
- In un-operated patients with large defects pulmonary vascular disease develops → shunt reversal and cyanosis (Eisenmenger's complex)

# Ventricular Septal Defect Anatomy

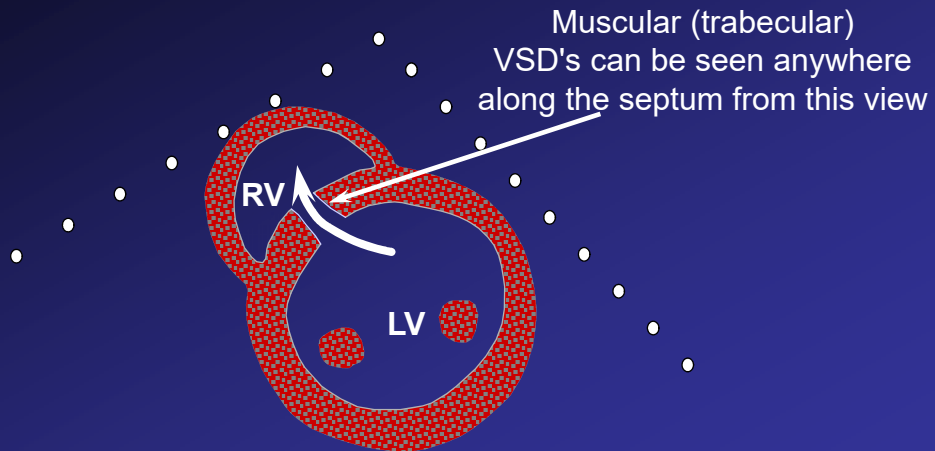


## Ventricular Septal Defect Parasternal Short Axis - Base



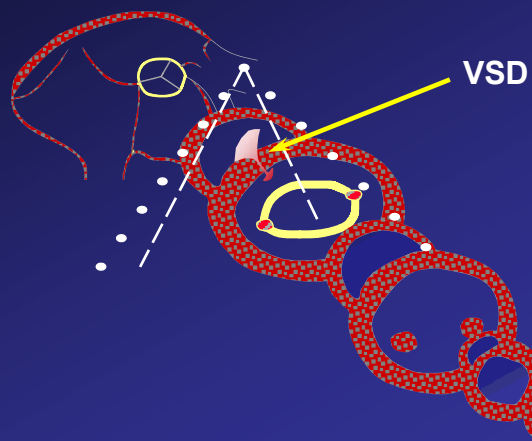
## Ventricular Septal Defect

Parasternal Short-Axis - Mid-Ventricle



## Ventricular Septal Defect

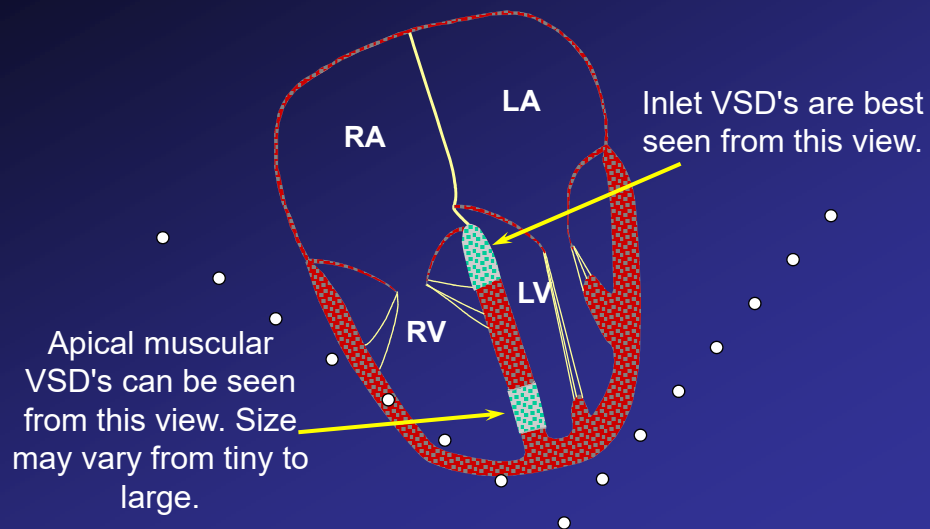
Parasternal Short-Axis "Sweep"





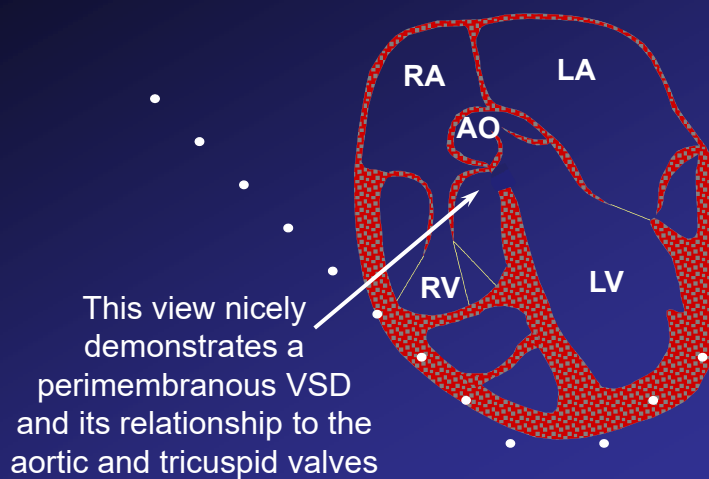
## Ventricular Septal Defect

### Apical Four- Chamber View

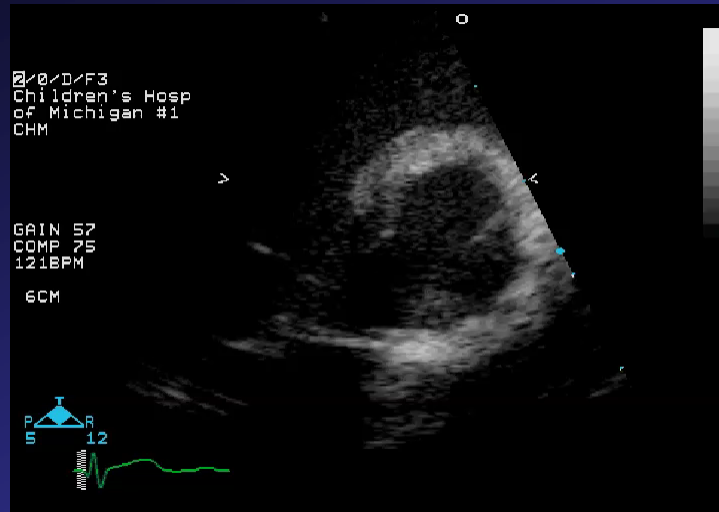


## Ventricular Septal Defect

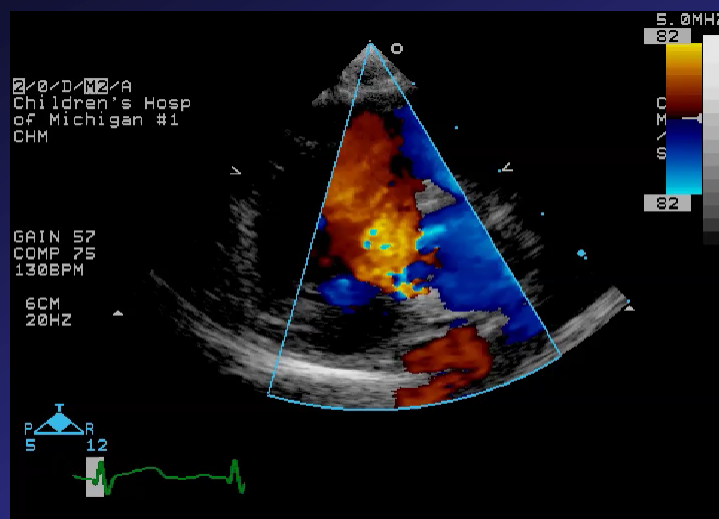
### Apical Five-Chamber View



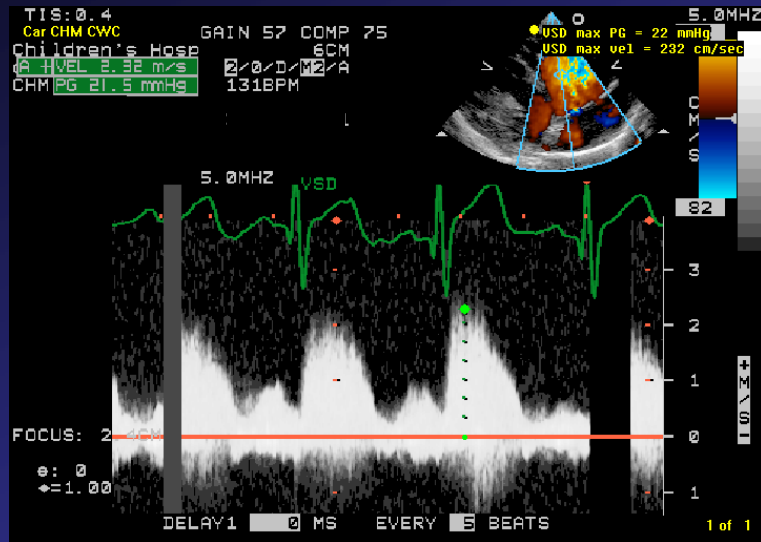
## Case 1- Review



## Case 1- Review

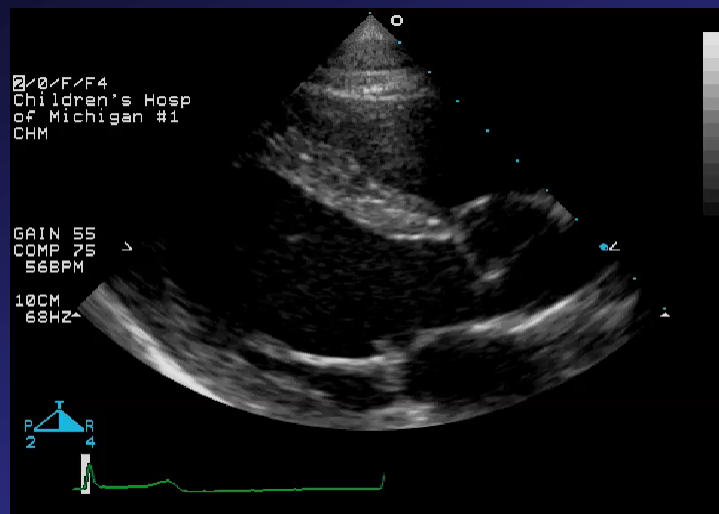


# Case 1- Review

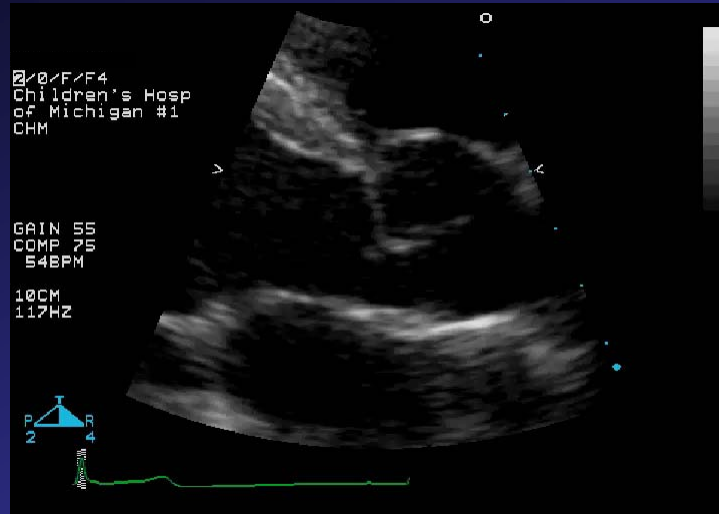


## Ventricular Septal Defect

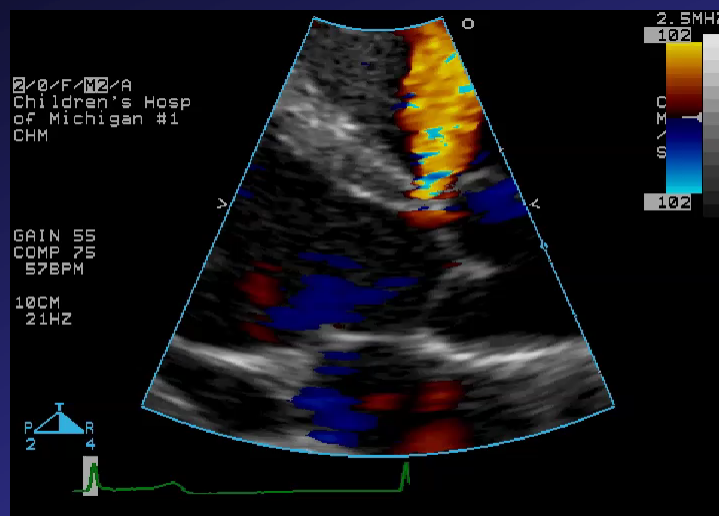
Case 2 – 8 y.o. with asymptomatic murmur



## Case 2 – 8 y.o. with asymptomatic murmur

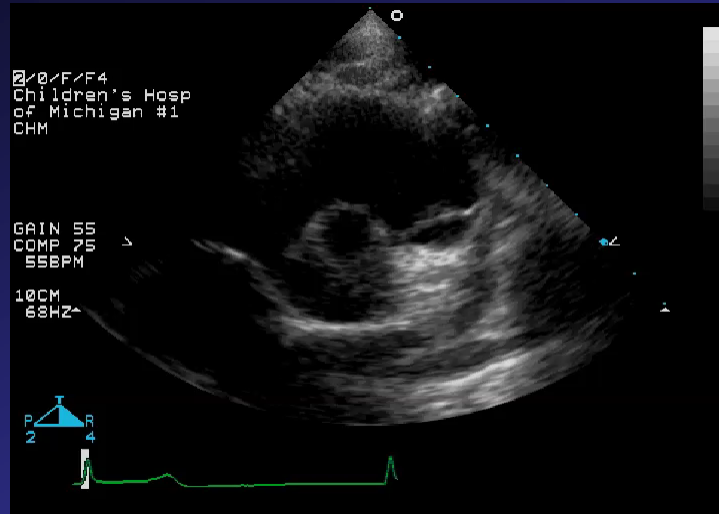


## Case 2 – 8 y.o. with asymptomatic murmur



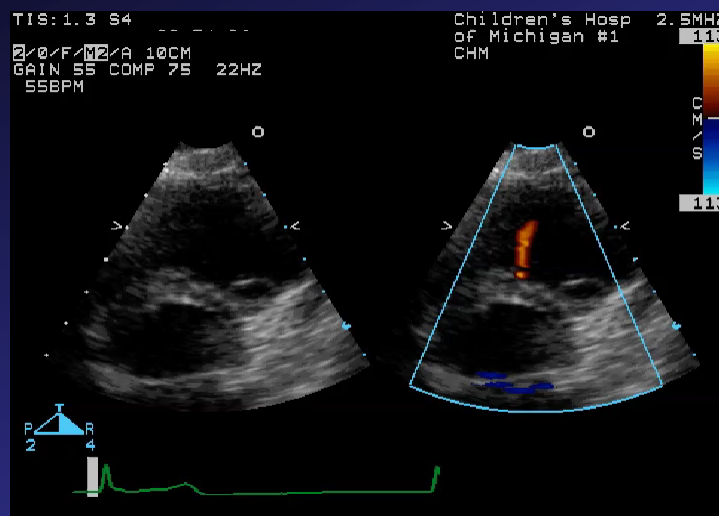
## Ventricular Septal Defect

Case 2 – 8 y.o. with asymptomatic murmur



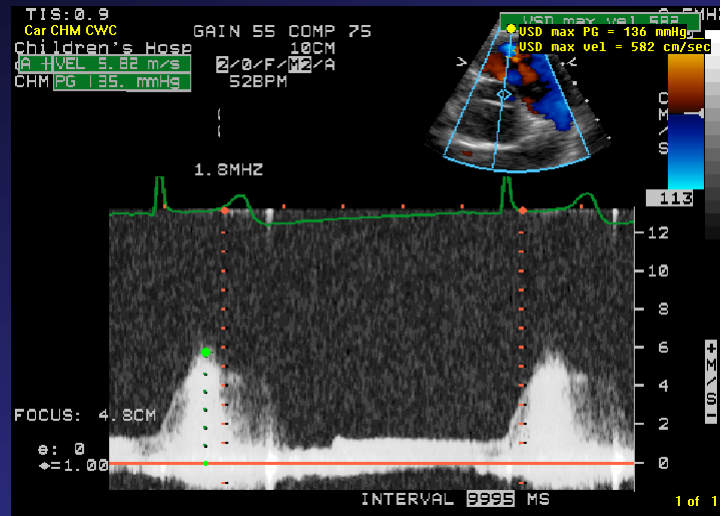
## Ventricular Septal Defect

Case 2 – 8 y.o. with asymptomatic murmur



## Ventricular Septal Defect

Case 2 – 8 y.o. with asymptomatic murmur



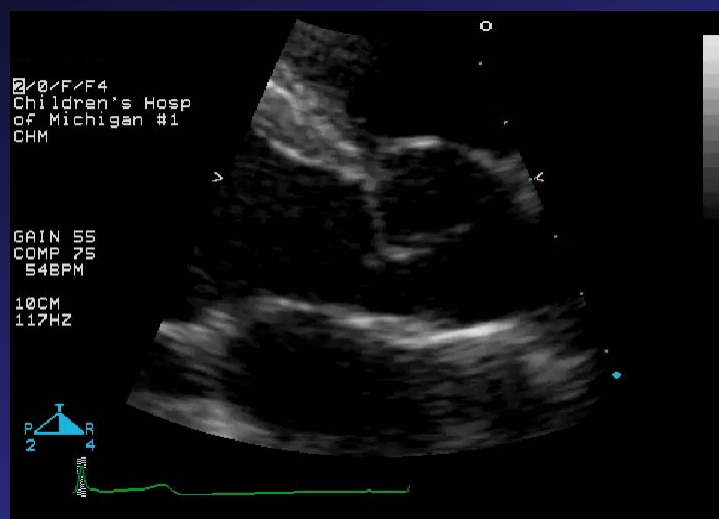
The defect shown in this example is:

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2. Sinus Venosus VSD
3. Perimembranous VSD
4. Inlet VSD
- ★ 5. Supracristal VSD

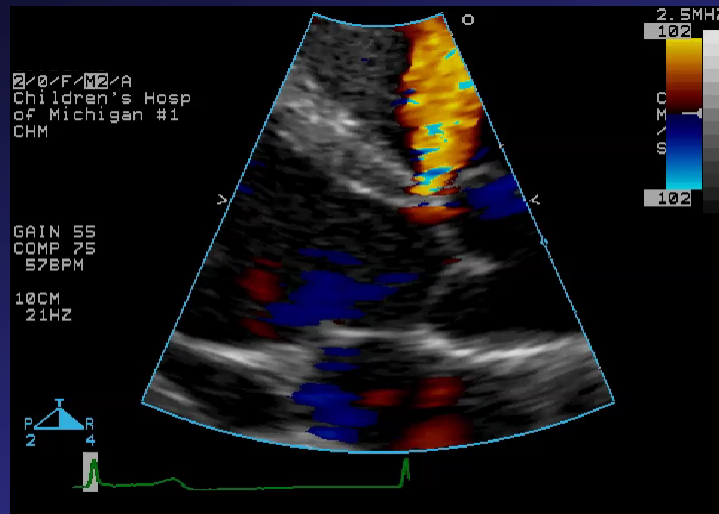
## Question 10 - A common complication of this defect is:

1. Pulmonary valve endocarditis
- ★ 2. Aortic regurgitation
3. Aortic dissection
4. Tricuspid regurgitation
5. Right ventricular enlargement

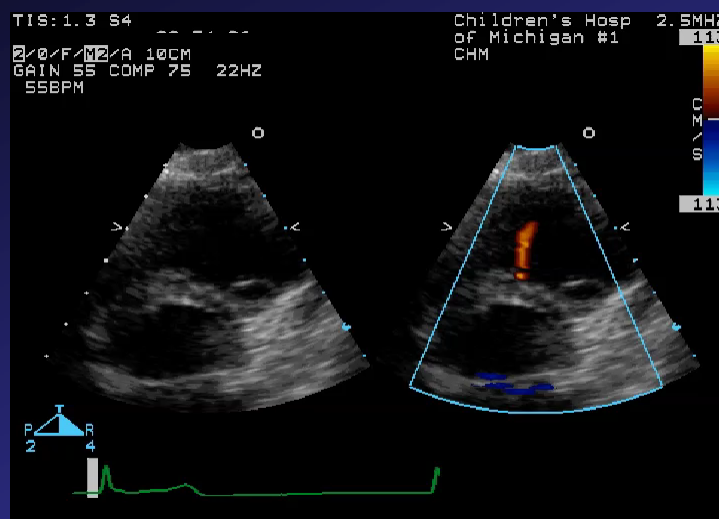
## Case 2- Review



## Case 2- Review



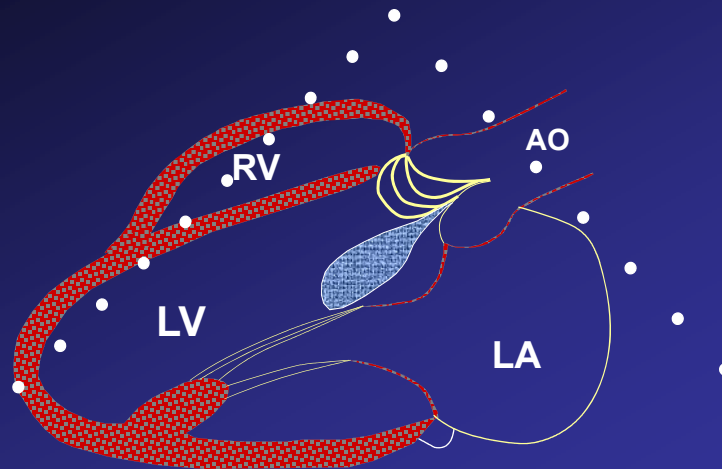
## Case 2- Review





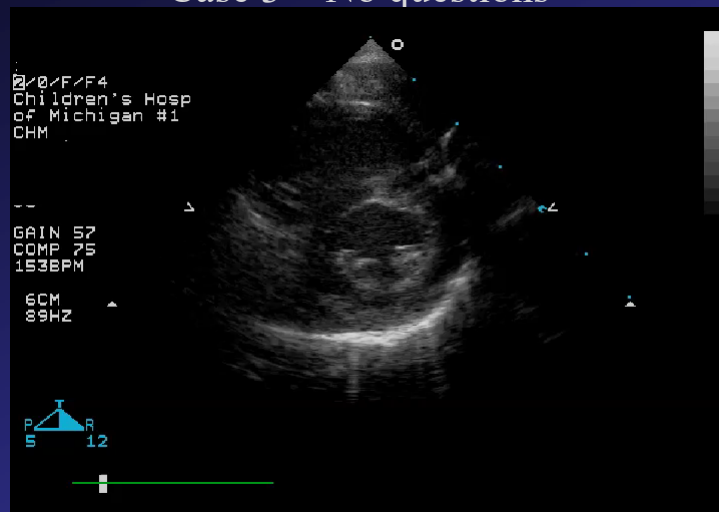
## Supracristal VSD

Aortic Cusp Prolapse



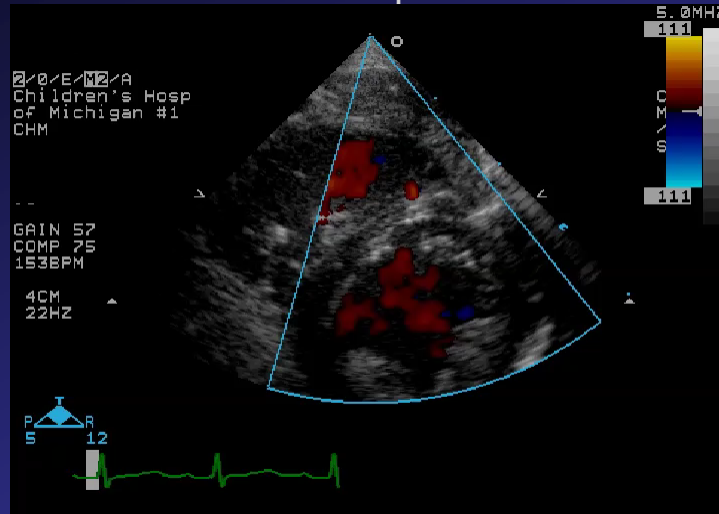
## Ventricular Septal Defect

Case 3 – No questions



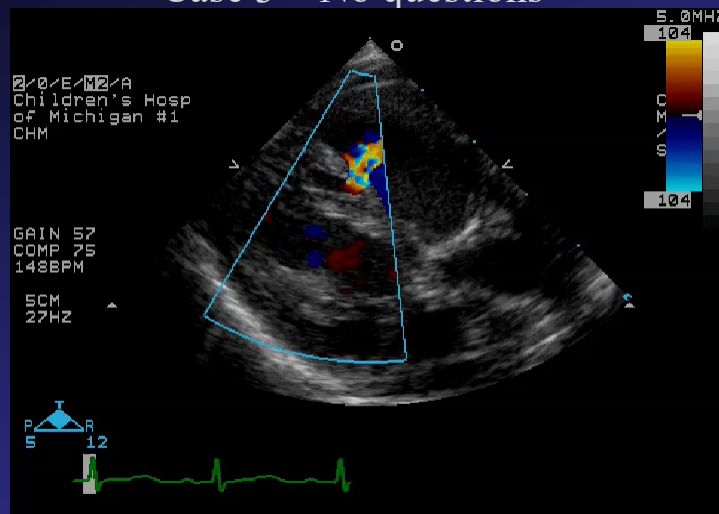
## Ventricular Septal Defect

Case 3 – No questions



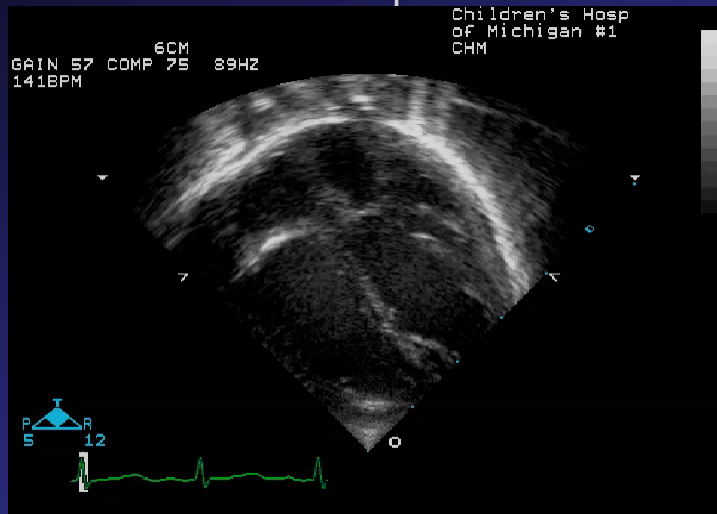
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Case 3 – No questions



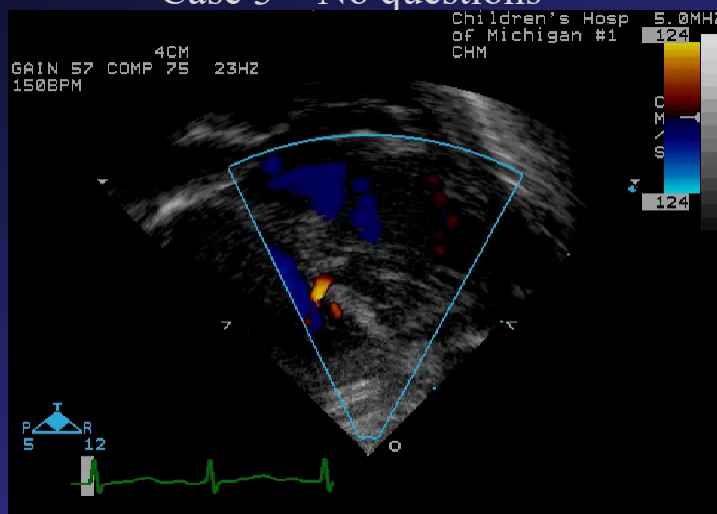
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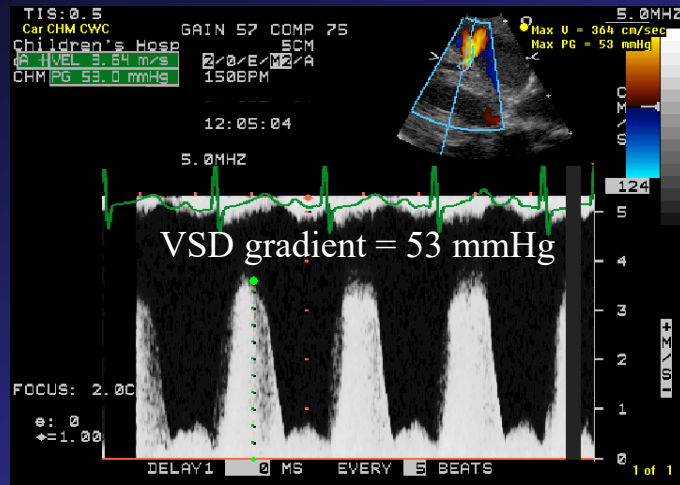
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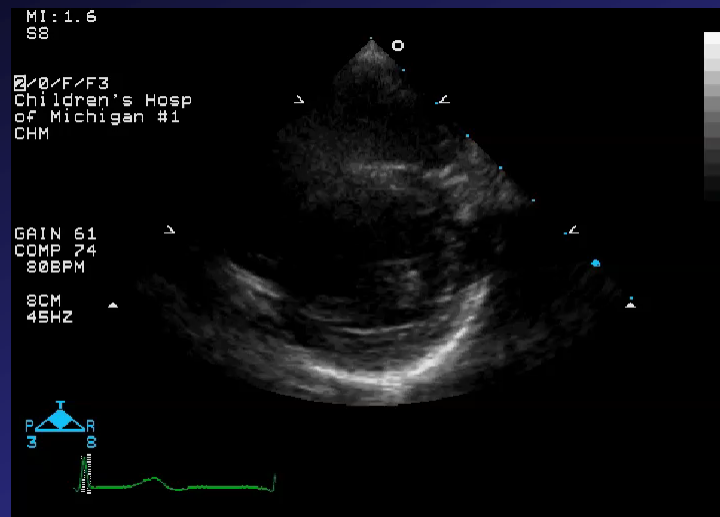
# Ventricular Septal Defect

## Case 3 – No questions

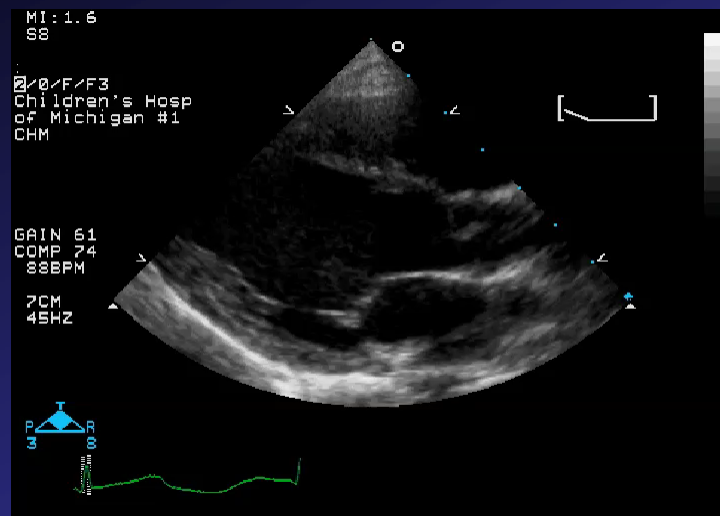


BP = 72/38 mmHg    RVSP = 72 – 53 = 19 mmHg

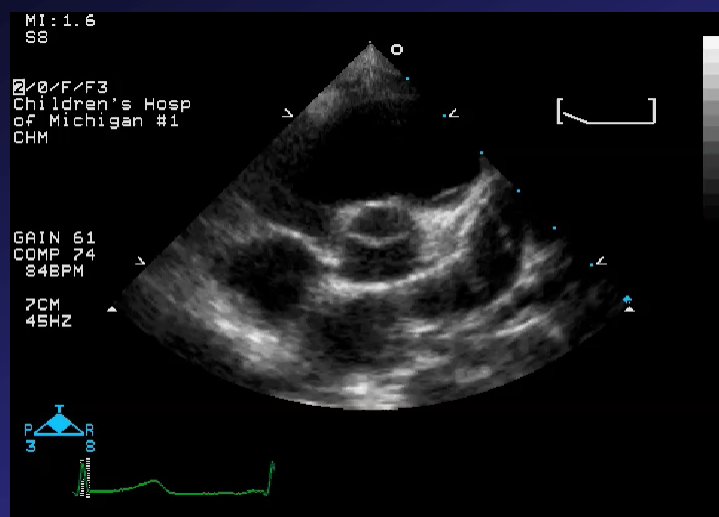
## Case 4 – 6 y.o. with continuous



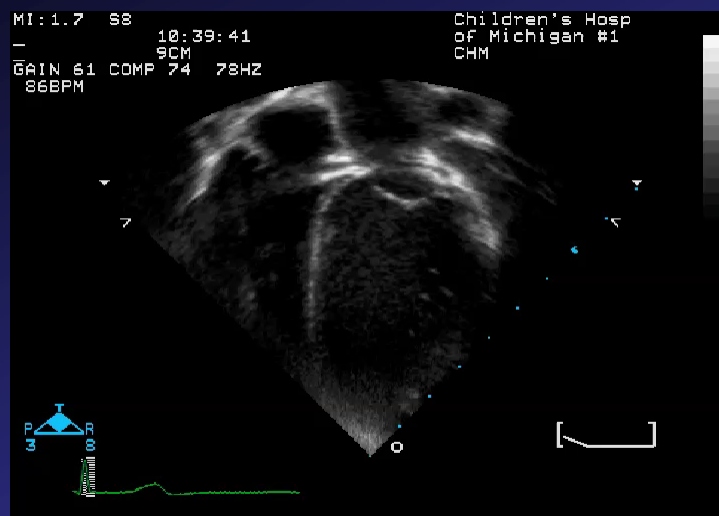
## Case 4 – 6 y.o. with continuous



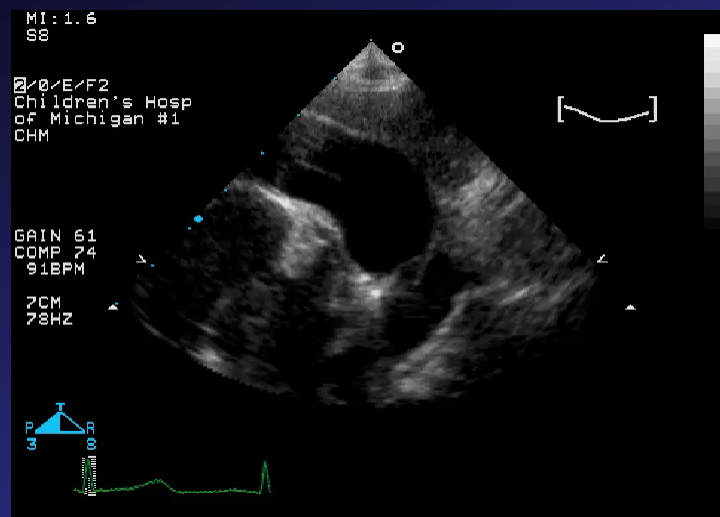
## Case 4 – 6 y.o. with continuous



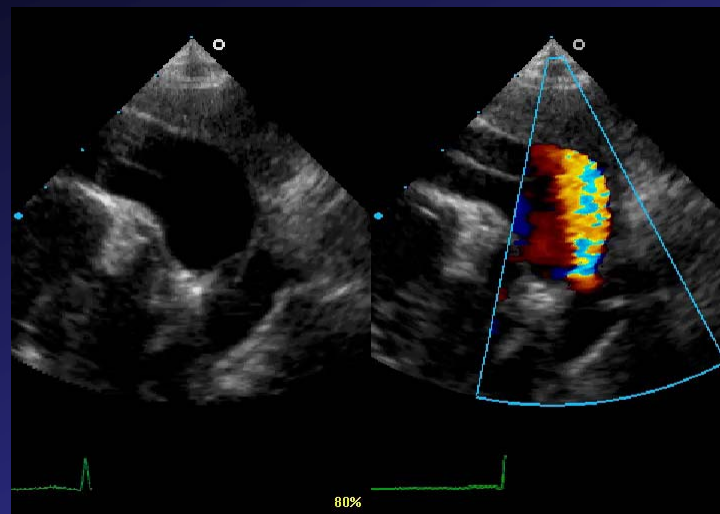
## Case 4 – 6 y.o. with continuous



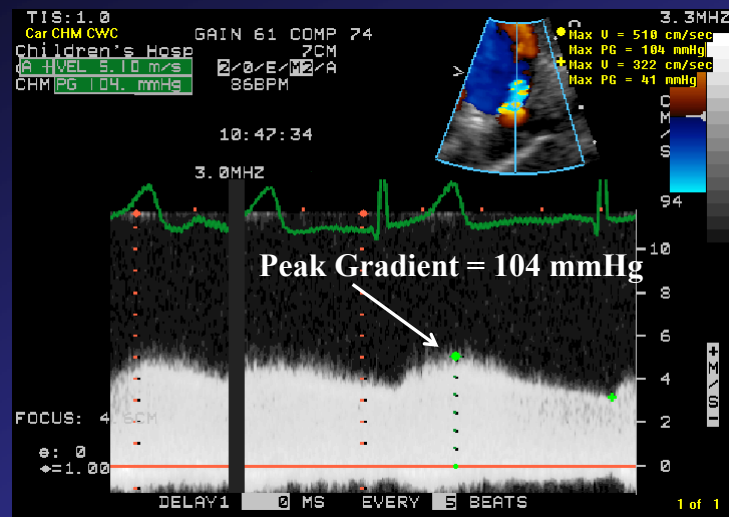
## Case 4 – 6 y.o. with continuous



## Case 4 – 6 y.o. with continuous



## Case 4 – 6 y.o. with continuous murmur



### The Doppler tracing in this case implies:

1. Severe pulmonary hypertension
2. Severe systemic hypertension
3. Severe coarctation of the aorta
- ★ 4. Normal pulmonary artery pressure
5. Severe pulmonary artery stenosis



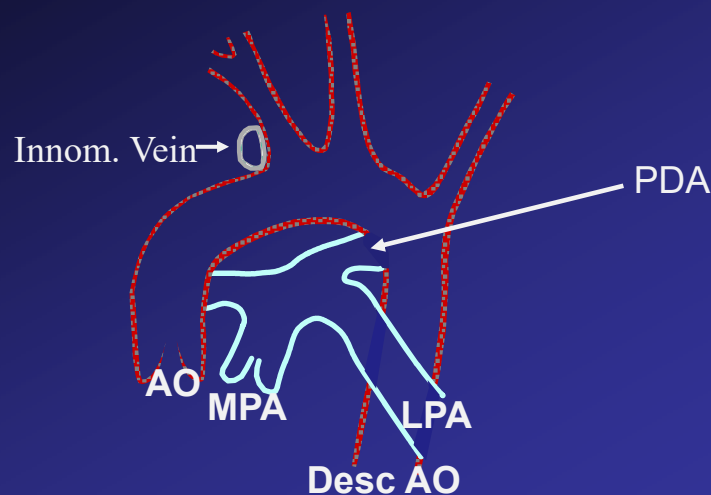
# Patent Ductus Arteriosus

## Clinical

- Continuous murmur in older patients
- Bounding pulses, wide pulse pressure, respiratory symptoms in neonates with a large PDA
- Large PDA will act much like a large VSD, producing pulmonary over-circulation and signs/symptoms of congestive heart failure
- A small PDA is generally hemodynamically insignificant but is at risk for endarteritis

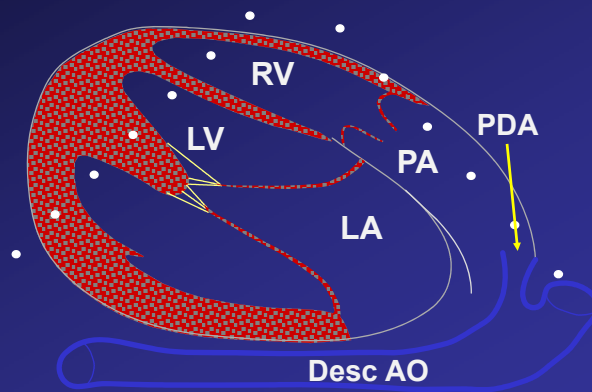
# Patent Ductus Arteriosus

## Anatomy

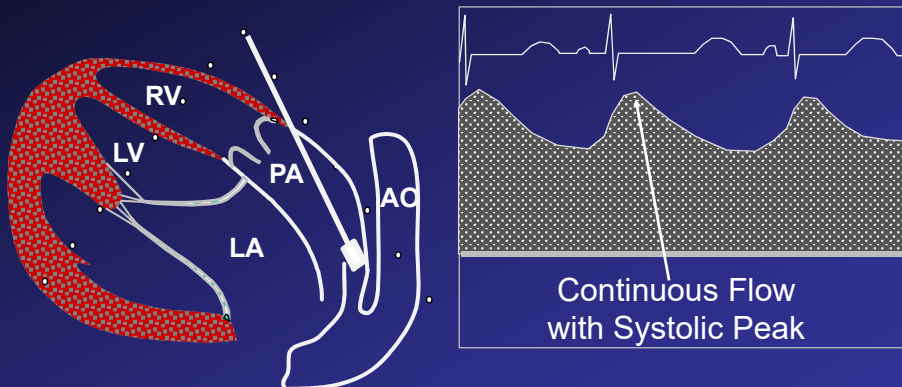


# Patent Ductus Arteriosus

Ductal View  
Parasternal Ductal View  
(High Left Parasternal)



# Patent Ductus Arteriosus Doppler Flow Pattern



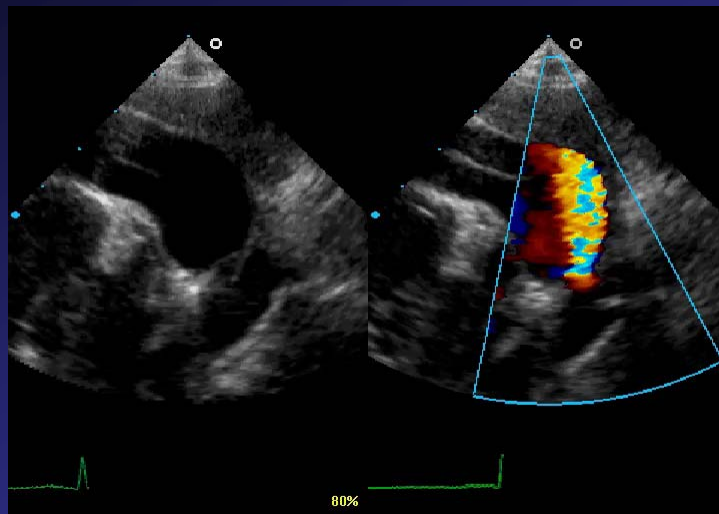
$$\text{Systolic PA pressure} = \text{SBP} - 4V_{\text{PDA}}^2$$

# Patent Ductus Arteriosus

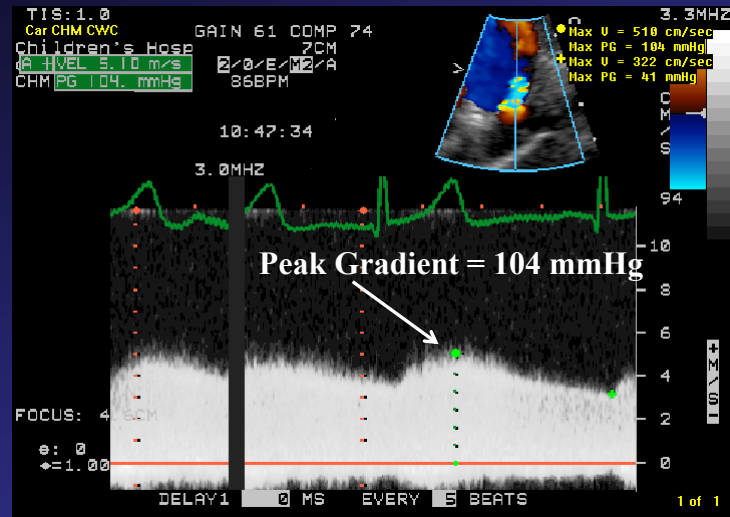
## Clinical management

- Large PDAs in preterm infants
  - Pharmacologic closure – indomethacin or ibuprofen
  - Surgical closure – left lateral thoracotomy
- Small PDA in older infants and children
  - Catheter closure – device or coil

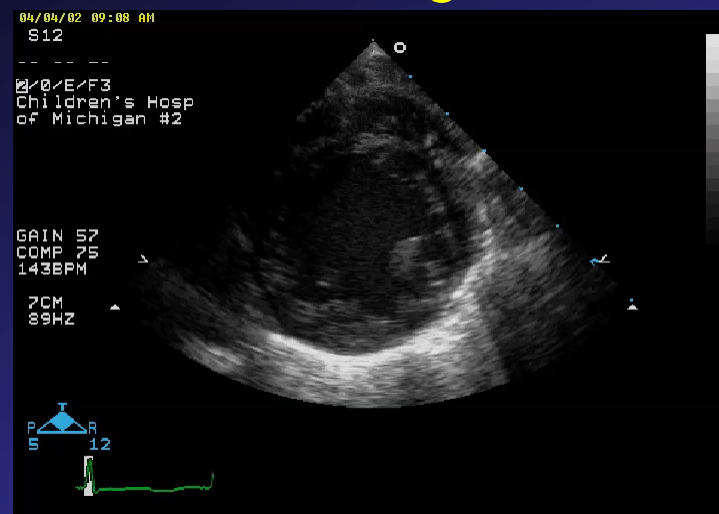
## Case 4 - Review



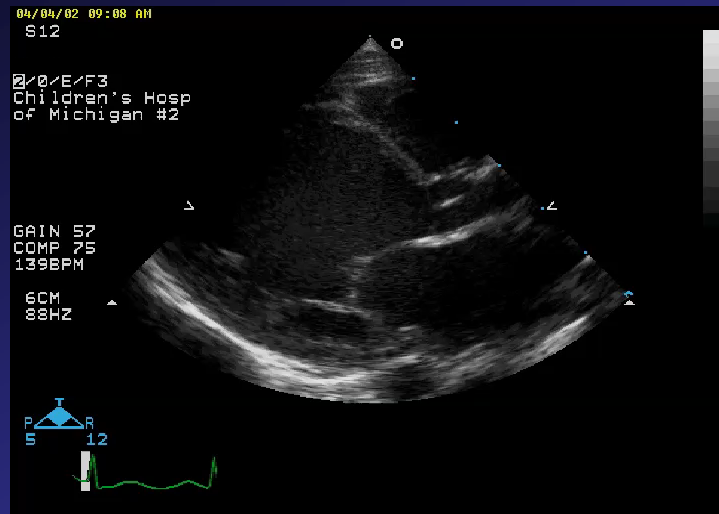
## Case 4 - Review



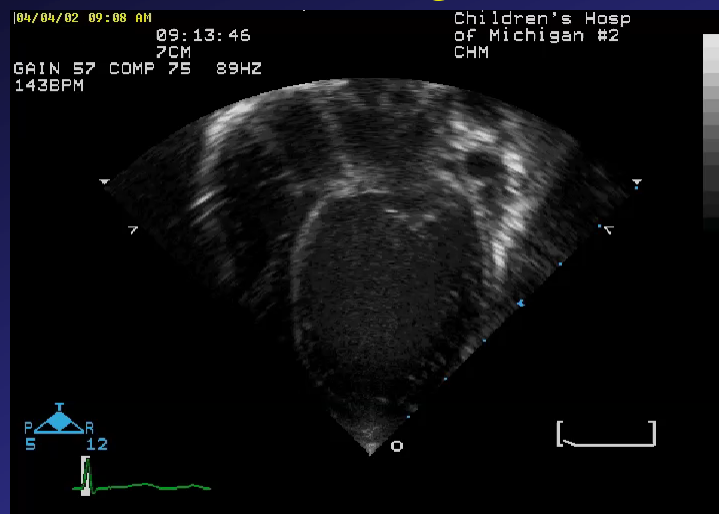
## Case 5 – Large PDA



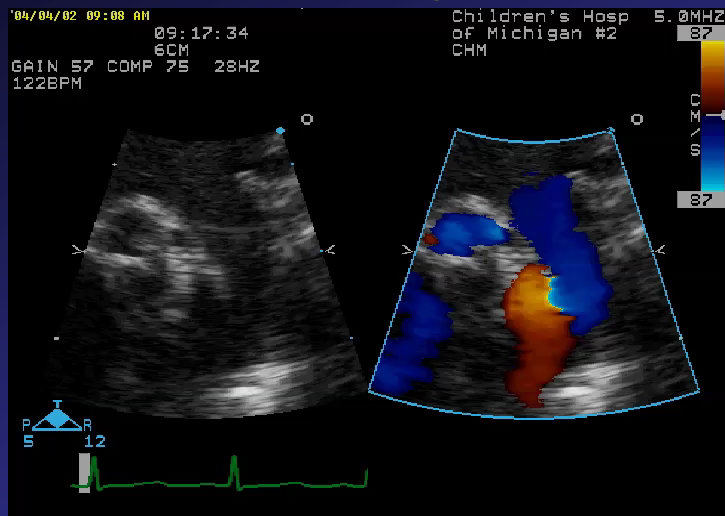
## Case 5 – Large PDA



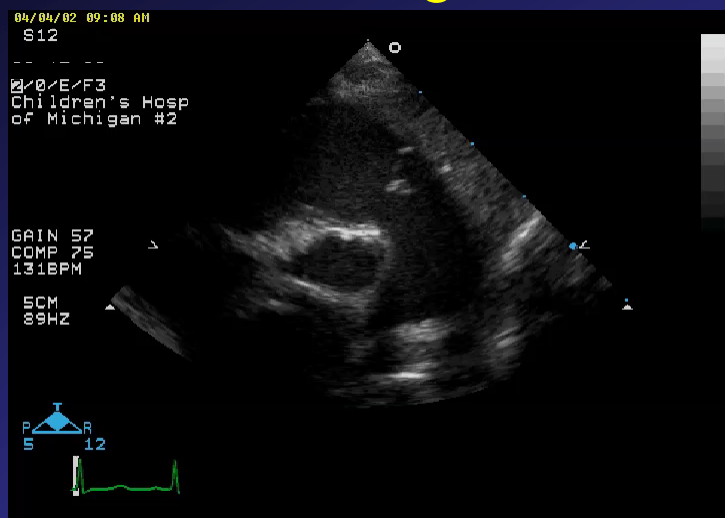
## Case 5 – Large PDA



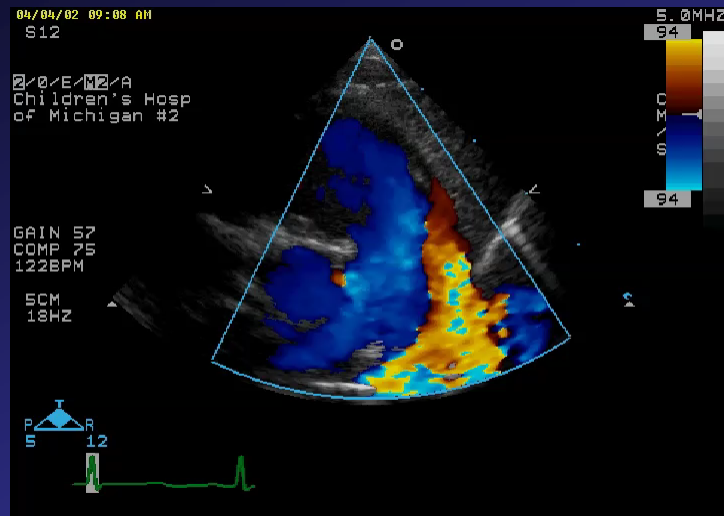
## Case 5 – Large PDA



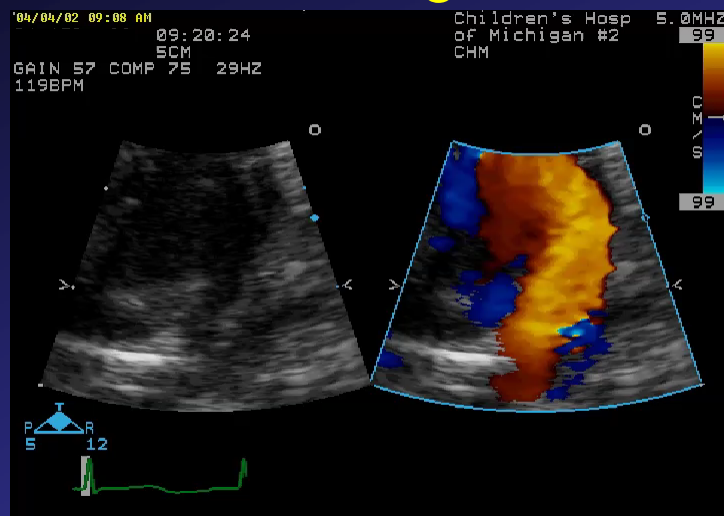
## Case 5 – Large PDA



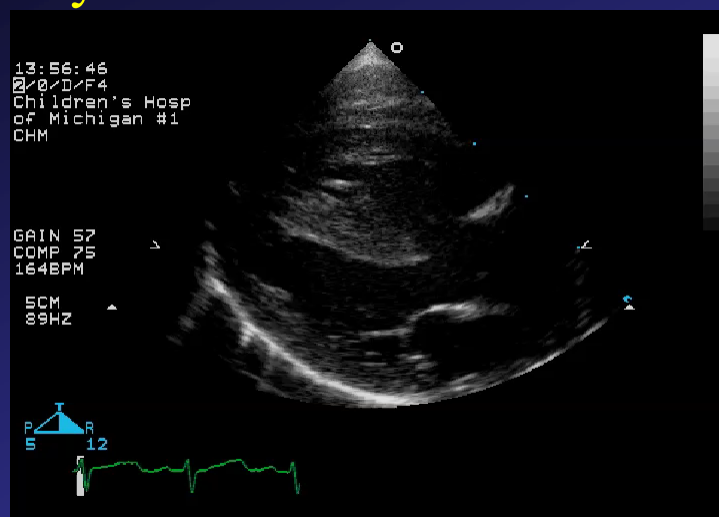
## Case 5 – Large PDA



## Case 5 – Large PDA

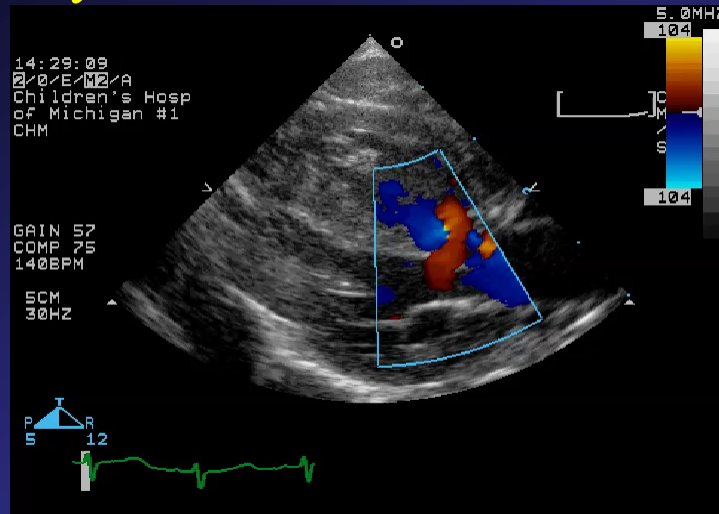


## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur

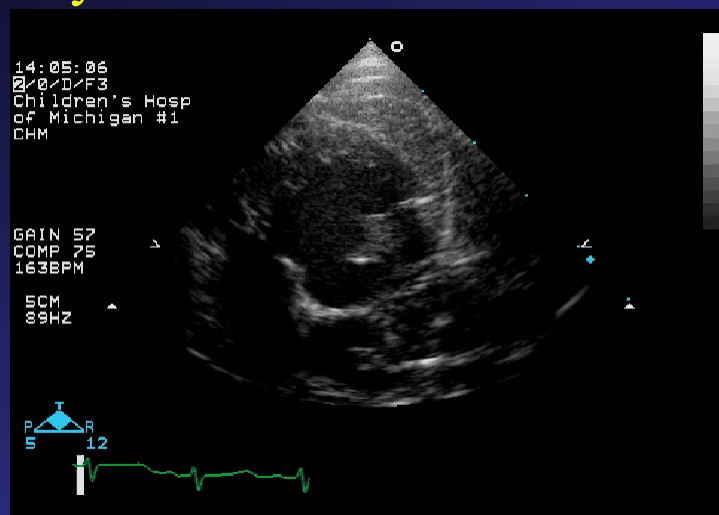




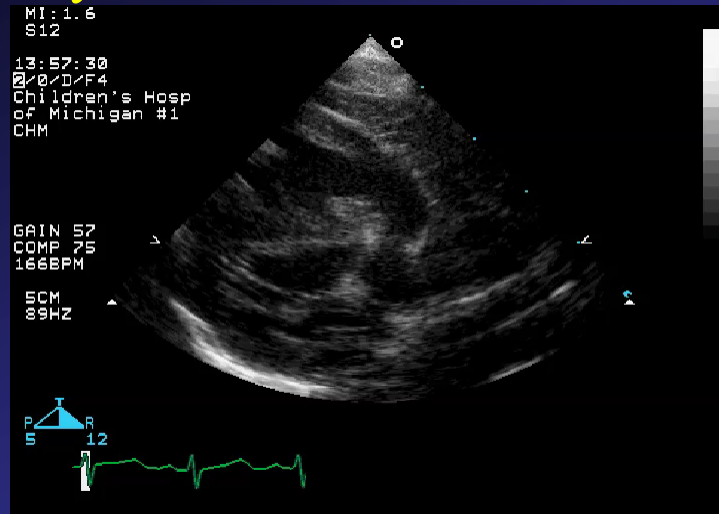
## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



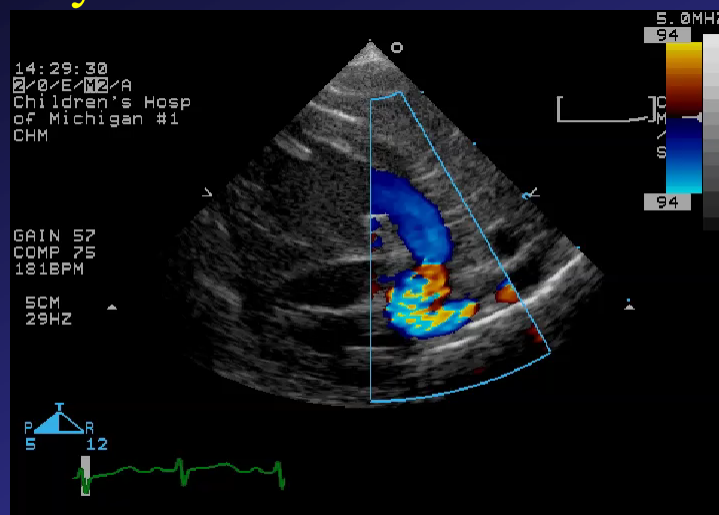
## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



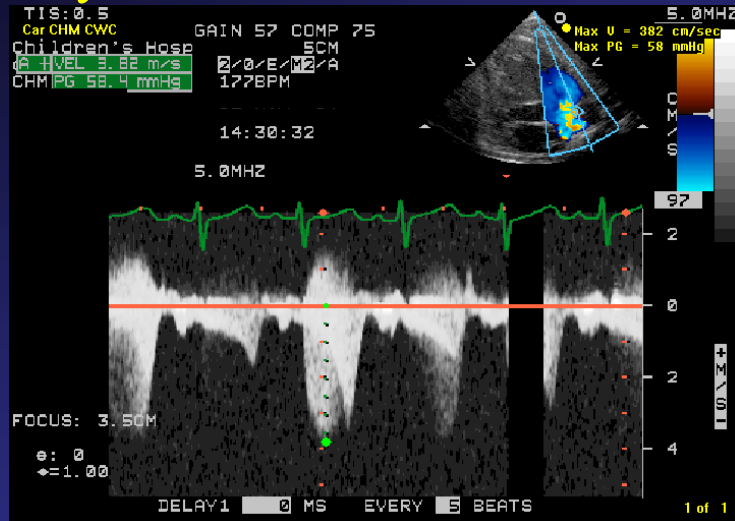
## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



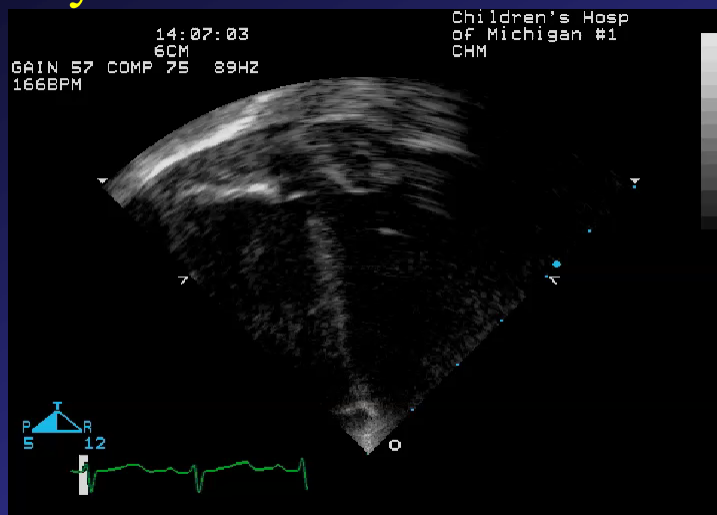
## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



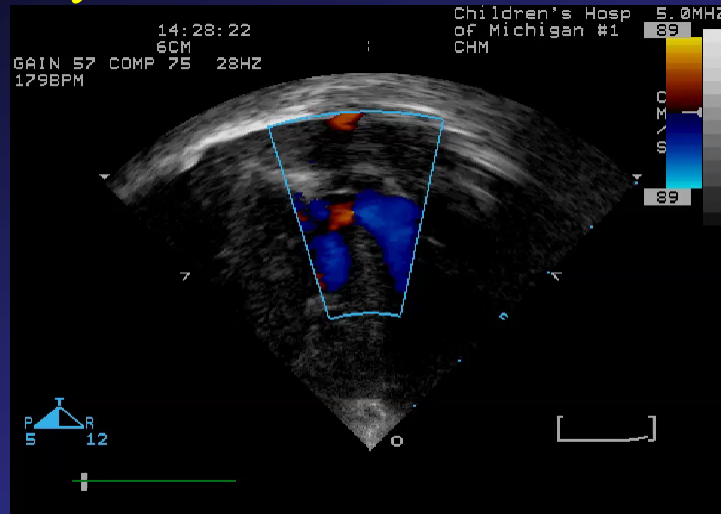
## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



## Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



The defect shown in this example is:

1. Single ventricle
2. Transposition of the great arteries
3. Perimembranous VSD
- ★ 4. Tetralogy of Fallot
5. Complete atrioventricular canal

# Tetralogy of Fallot

## Background

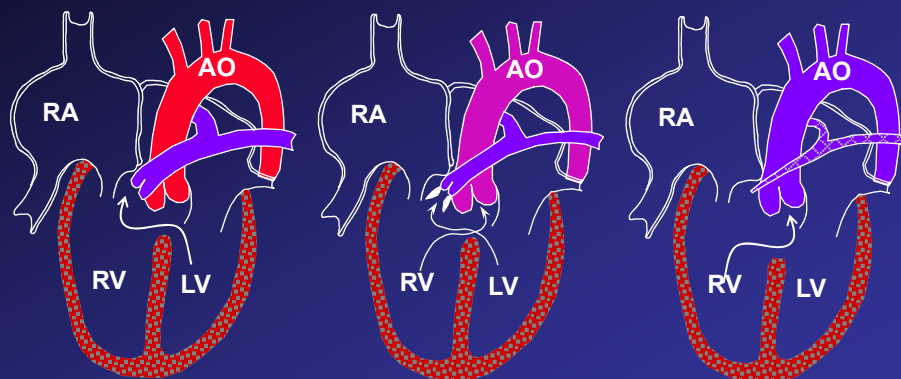
- Accounts for 10-13% of congenital heart disease
- Most common cyanotic CHD
- Usually present as asymptomatic murmur
  - Cyanosis usually develops/progresses with time
- Anatomy
  - Ventricular Septal Defect
  - Overriding Aorta
  - RV outflow obstruction
  - RV hypertrophy



Conal Septum Malalignment

# Tetralogy of Fallot

## Anatomic Variables



"Pink Tetralogy"  
Mild Pulmonary Stenosis

Classic Tetralogy

Severe Tetralogy or  
Pulmonary Atresia

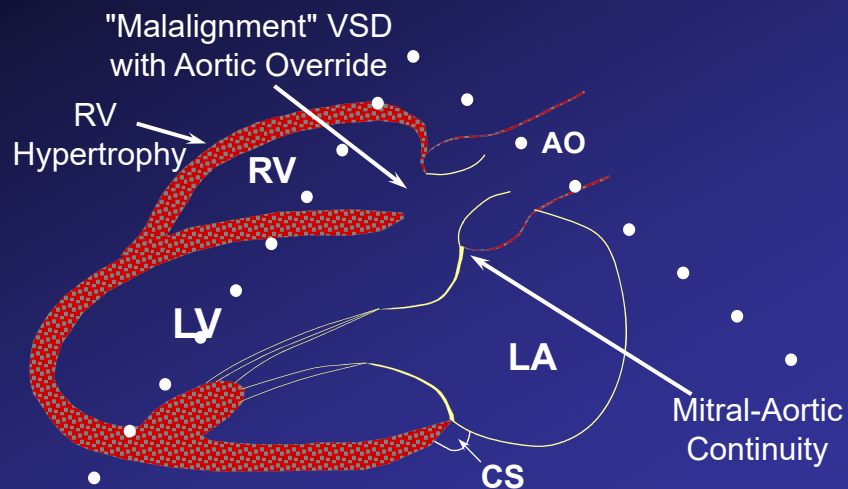
# Tetralogy of Fallot

## Associated Anomalies

- Valvular pulmonary stenosis - 50-60%
- Right aortic arch - 25%
- Atrial septal defect - 15%
- Coronary anomalies - 5%
- Muscular VSD - 2%

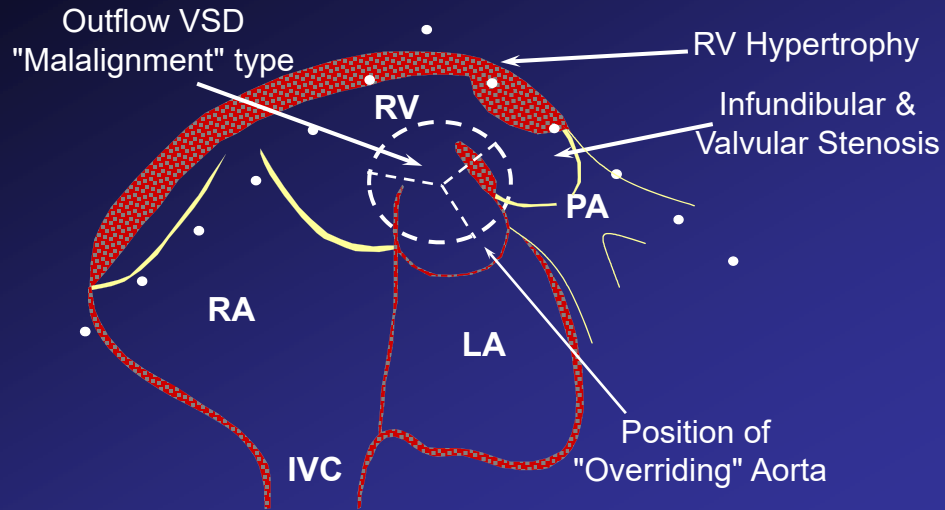
# Tetralogy of Fallot

## Parasternal Long Axis View



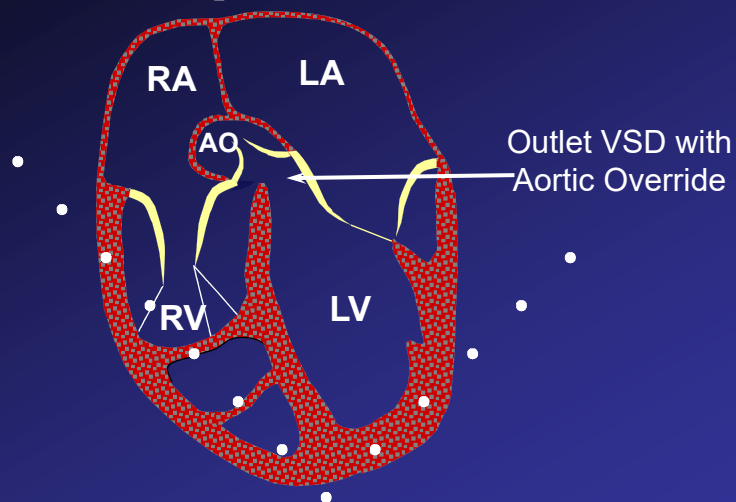
## Tetralogy of Fallot

Parasternal Short Axis - Base



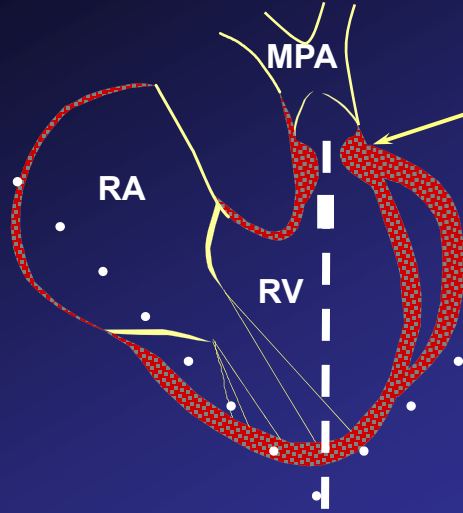
## Tetralogy of Fallot

Apical Five-Chamber



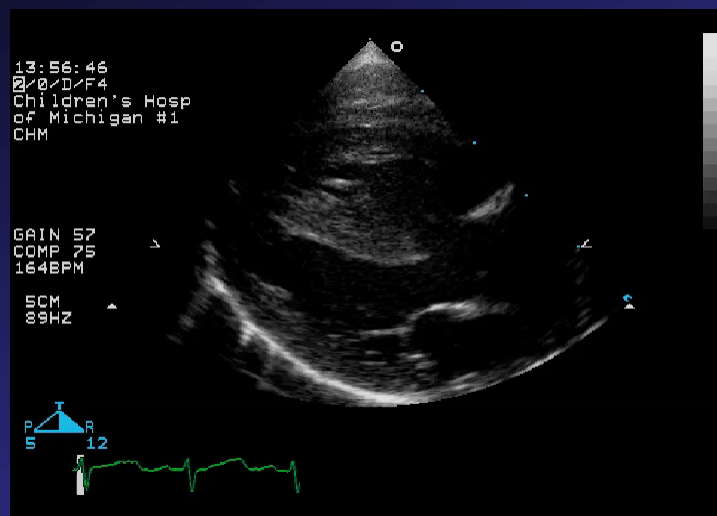
# Tetralogy of Fallot

## Subcostal RV Inflow/Outflow View



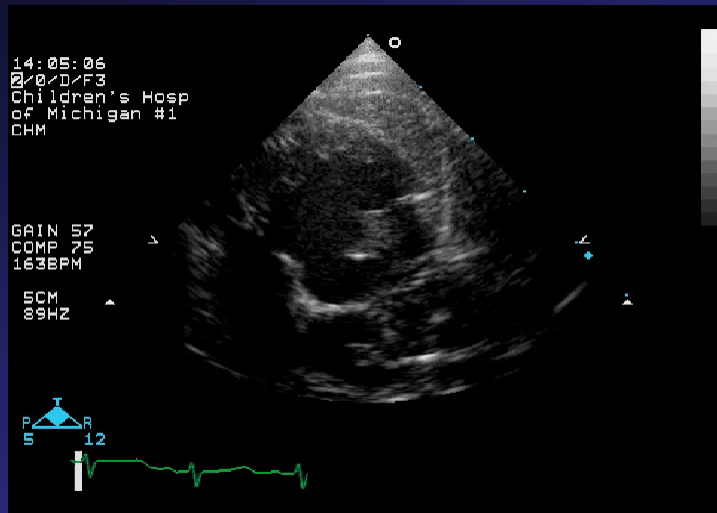
The right ventricular outflow obstruction is seen in this view. This is often the best angle for Doppler interrogation of the RVOT

## Case 6 - Review

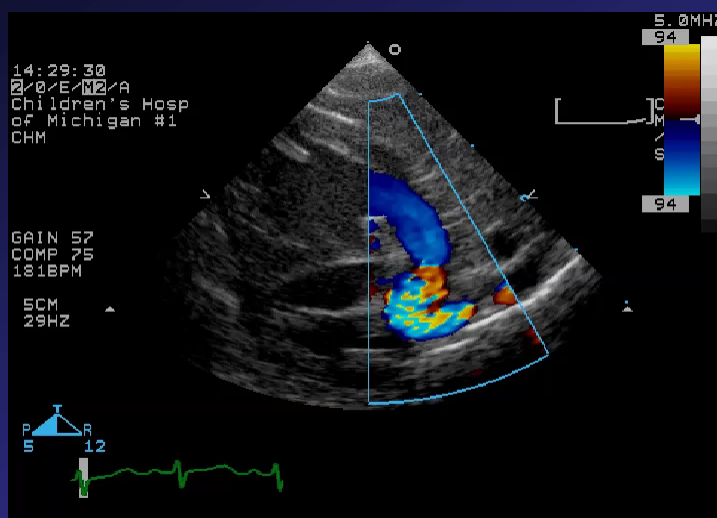




## Case 6 - Review



## Case 6 - Review

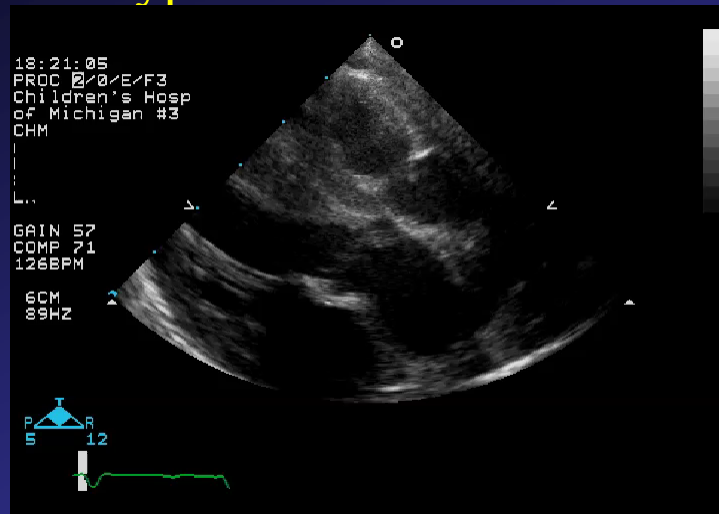


# Tetralogy of Fallot

## Surgical Intervention

- Timing – usually during first 6 months
- VSD closure, relief of RVOTO obstruction
- Many repairs require a trans annular RV outflow patch with results in chronic severe pulmonary regurgitation
  - Likely need for late pulmonary valve replacement
- Rastelli type repair (VSD closure + RV to pulmonary artery conduit) may be required for complex anatomy – pulmonary atresia, coronary anomalies

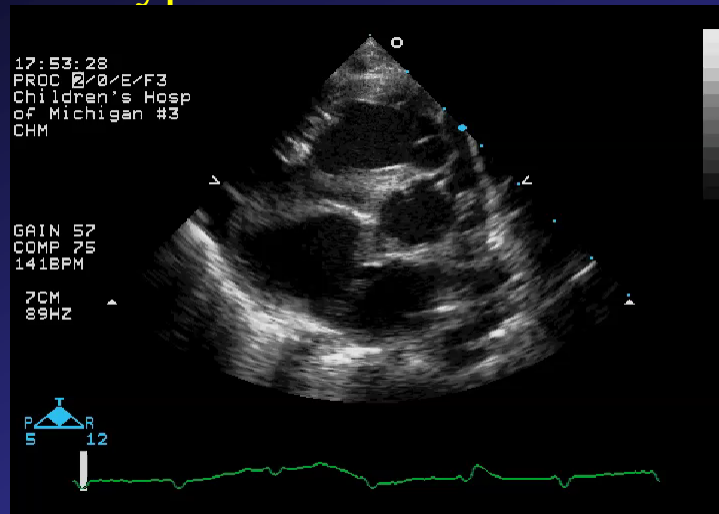
## Case 7 – 1 day old infant with tachypnea and SaO<sub>2</sub> of 76%



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## Case 7 – 1 day old infant with tachypnea and SaO<sub>2</sub> of 76%



## What congenital heart defect is shown:

1. Perimembranous VSD
2. Truncus arteriosus
3. Corrected transposition of the great arteries (L-TGA)
- ★ 4. Complete transposition of the great arteries (D-TGA)
5. Tetralogy of Fallot

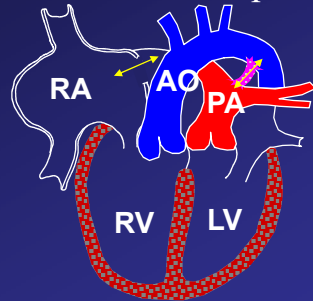
## Which of the following is the preferred surgical palliation of this defect?

1. Rastelli operation
2. Mustard operation
- ★ 3. Jatene operation
4. Konno operation
5. Fontan operation

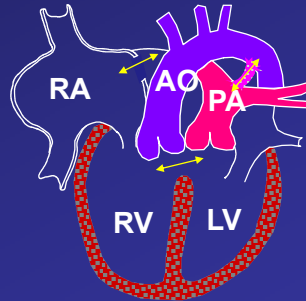
## D-Transposition of the Great Arteries

- Most common cyanotic CHD presenting in the newborn nursery
- 4-8% of CHD
- Very high mortality without intervention (90% at 1 year of life)

D-TGA Intact Septum



D-TGA w/ VSD



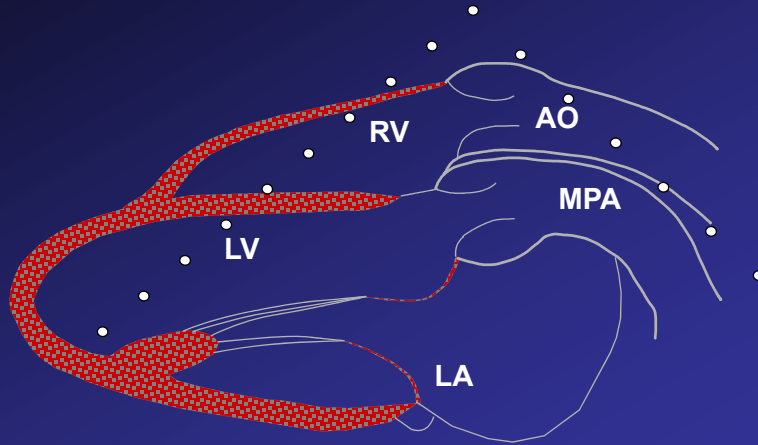
## D-Transposition of the Great Arteries

### Associated Anomalies

- VSD 40-45%
- Coronary anomalies ~ 40%
- Pulmonary stenosis (valve or sub valve) - 25%
- ASD
- PDA
- Coarctation - 5%

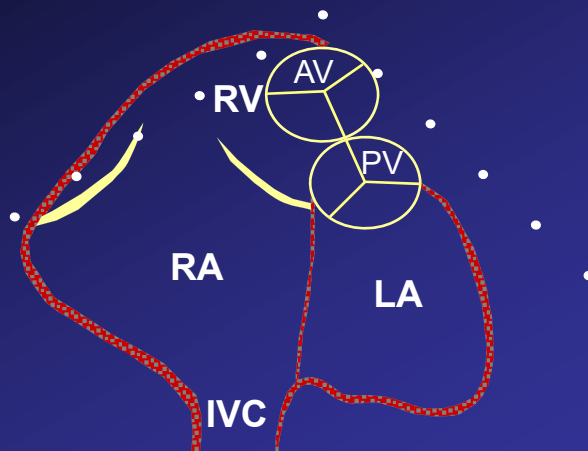
## D-Transposition of the Great Arteries

Parasternal Long Axis View



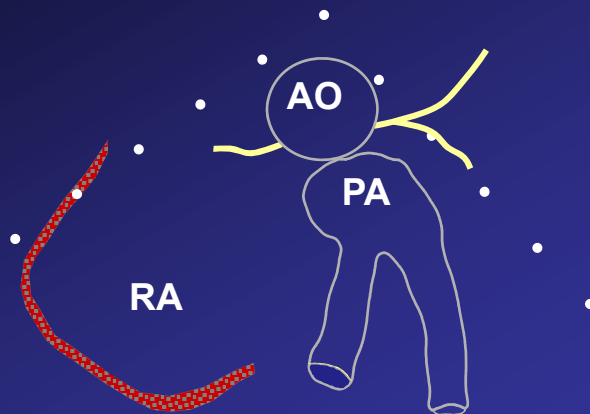
## D-Transposition of the Great Arteries

Parasternal Short Axis - Base

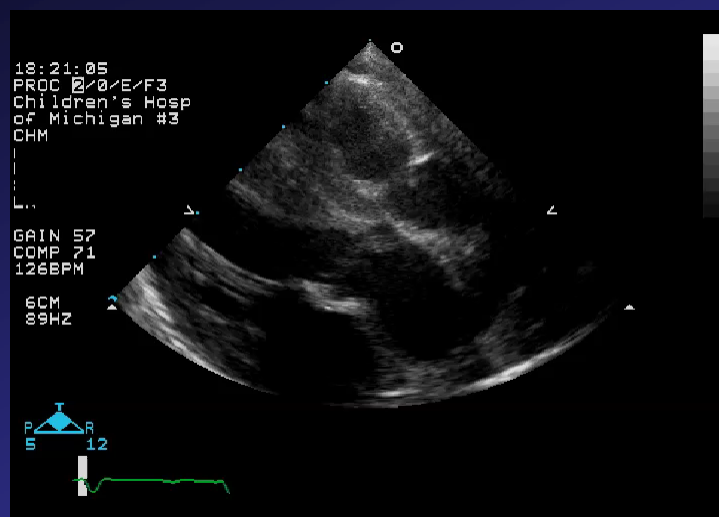


# D-Transposition of the Great Arteries

High Parasternal Short Axis - Base



## Case 7 – Review

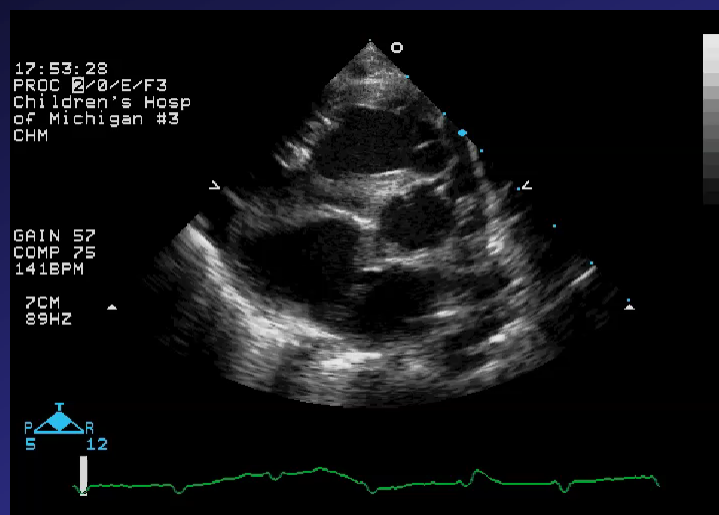




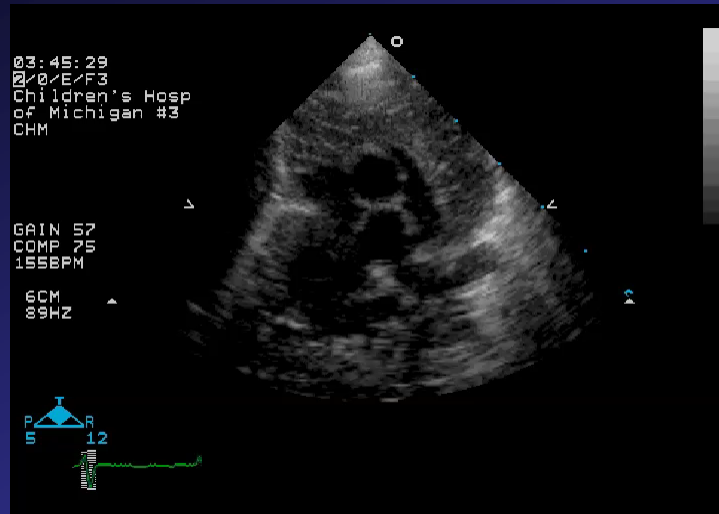
## Case 7 – Review



## Case 7 – Review



## Case 7 – Review



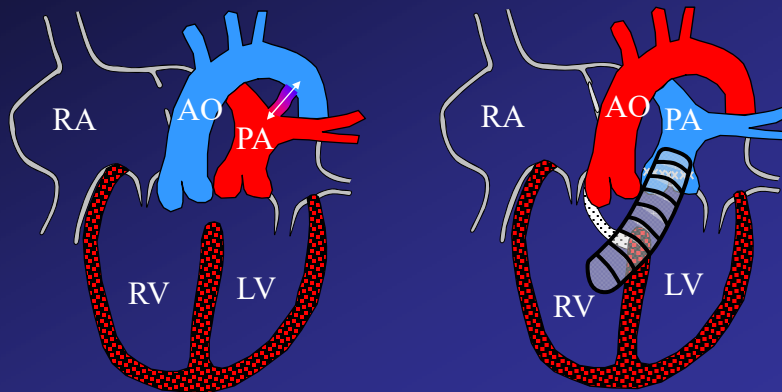
## D-Transposition of the Great Arteries

### Interventions

- Atrial balloon septostomy (Rashkind)
  - ♥ Increases saturation by improving atrial mixing
- Arterial switch (Jatene)
  - ♥ Performed within the first 1-2 weeks of life
- Atrial switch (Mustard/Senning)
  - ♥ Has been largely abandoned
- VSD closure/ RV-PA conduit (Rastelli)
  - ♥ Performed within the first few months of life
  - ♥ Used in the setting of d-TGA with PS/sub-PS
  - ♥ Requires conduit replacement/ future surgery

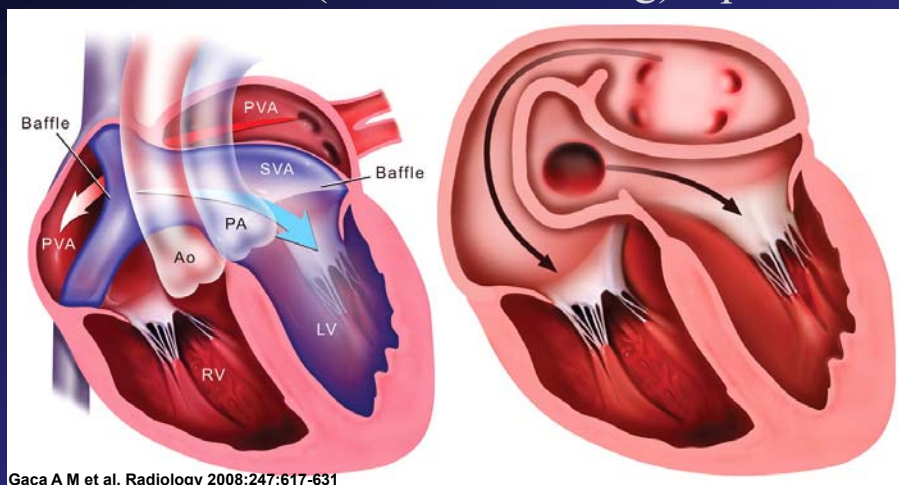
## D-TGA

### Intervention-Rastelli Procedure



## D-TGA

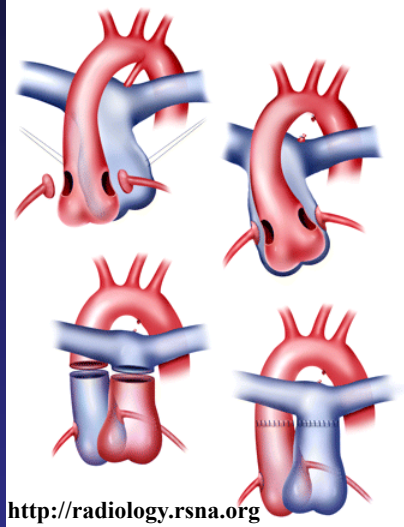
### Atrial Switch (Mustard/Senning) Operation



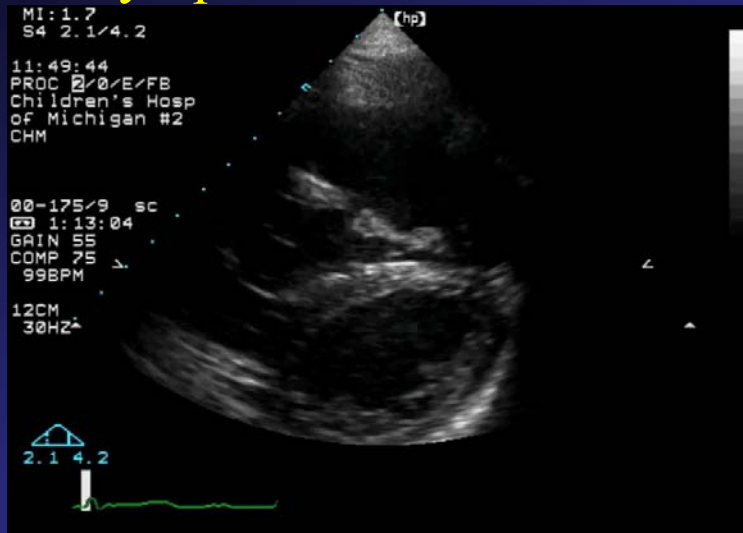
Gaca A M et al. Radiology 2008;247:617-631

## TGA

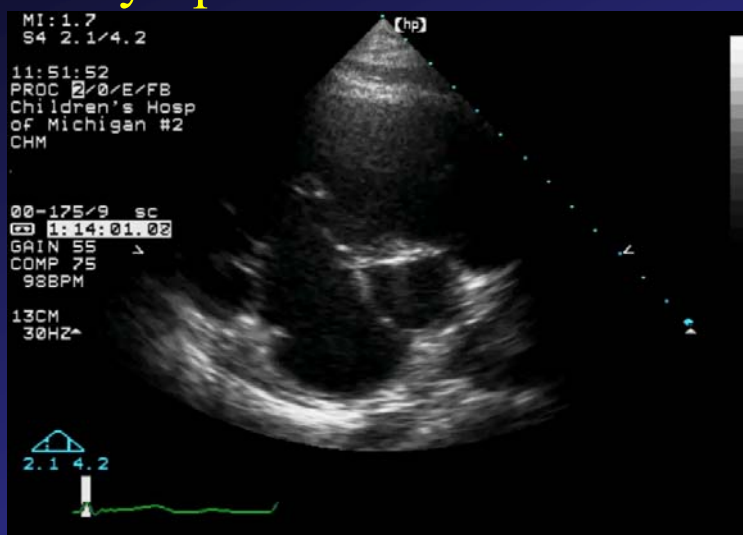
### Jatene Arterial Switch Operation



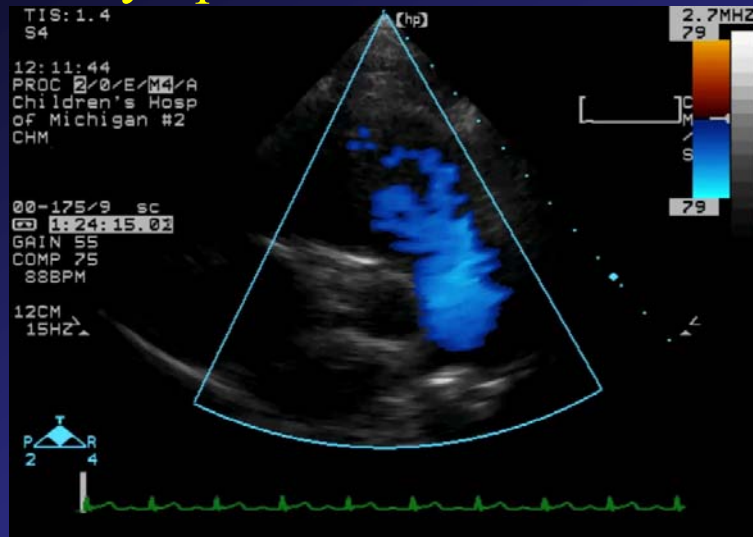
## Case 8 – 12 year old with asymptomatic murmur



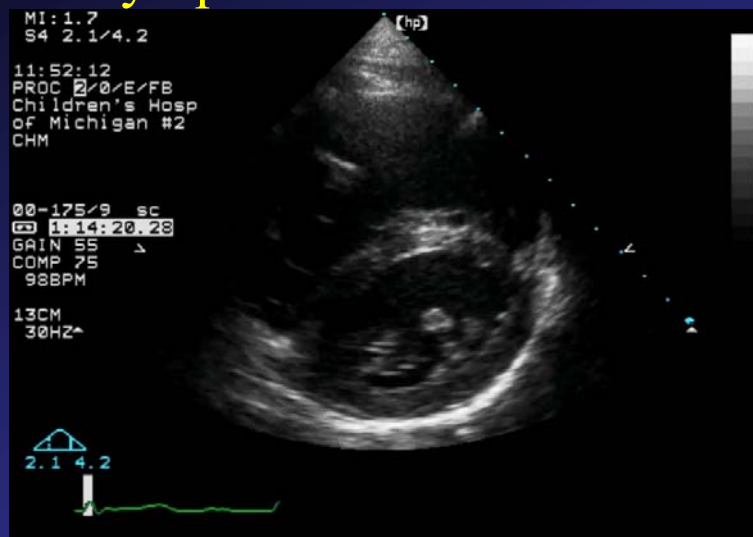
## Case 8 – 12 year old with asymptomatic murmur



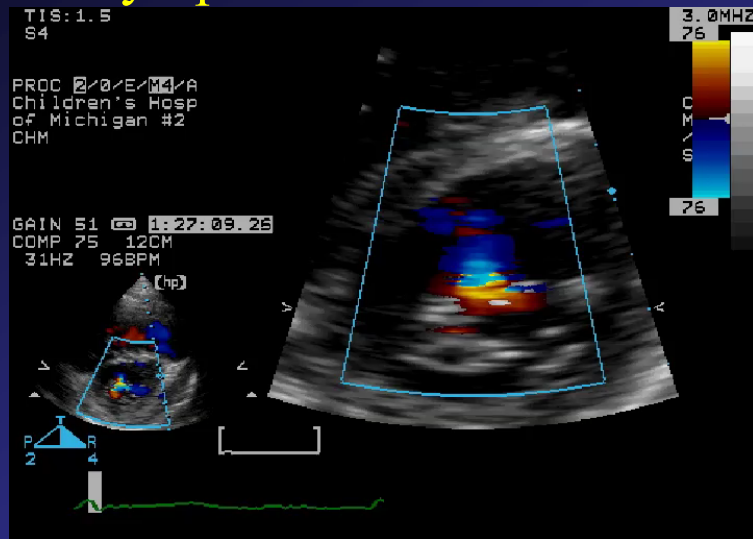
## Case 8 – 12 year old with asymptomatic murmur



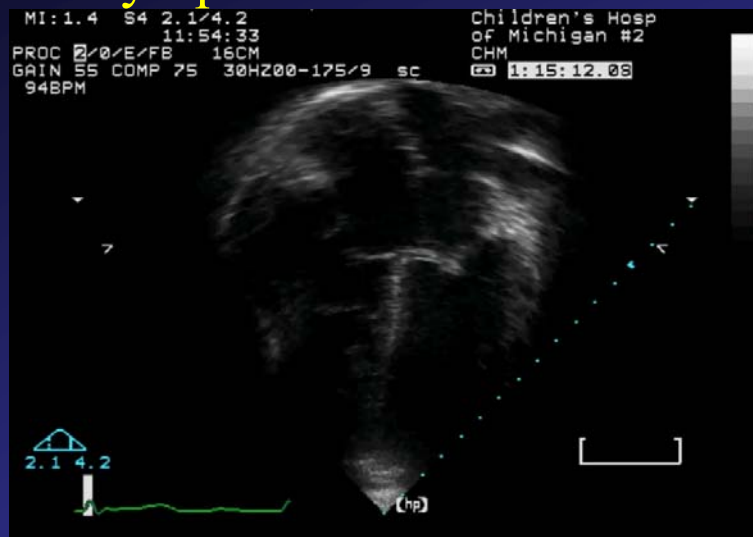
## Case 8 – 12 year old with asymptomatic murmur



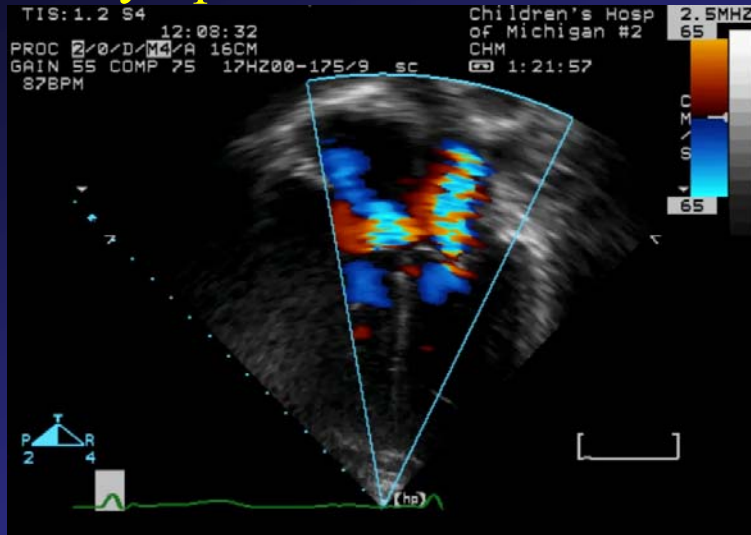
## Case 8 – 12 year old with asymptomatic murmur



## Case 8 – 12 year old with asymptomatic murmur



## Case 8 – 12 year old with asymptomatic murmur



The defect shown in this example is:

1. Secundum ASD
2. Sinus Venosus ASD
3. Perimembranous ASD
- ★ 4. Primum ASD
5. Coronary sinus ASD

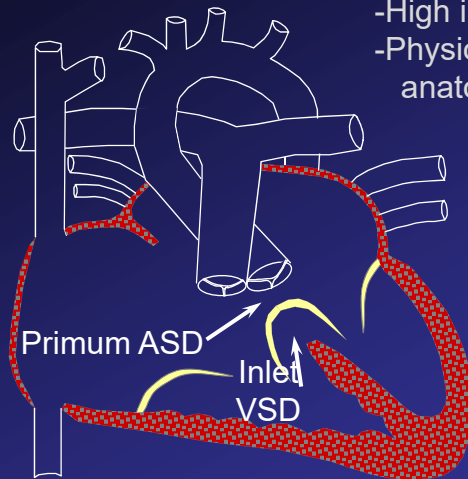


A common associated defect with this anomaly, shown in this case, is:

1. Bicuspid aortic valve
2. Perimembranous VSD
3. Patent ductus arteriosus
4. Coarctation of the aorta
- ★ 5. Cleft mitral valve

## Atrioventricular Septal Defects

- 3-5% of CHD
- High incidence in Down Syndrome
- Physiology depends on which anatomic defects are present



### Complete AVSD

1. Primum ASD
2. Inlet VSD
3. Common AV Valve

### Partial AVSD

1. Primum ASD
2. No VSD
3. Cleft Mitral Valve

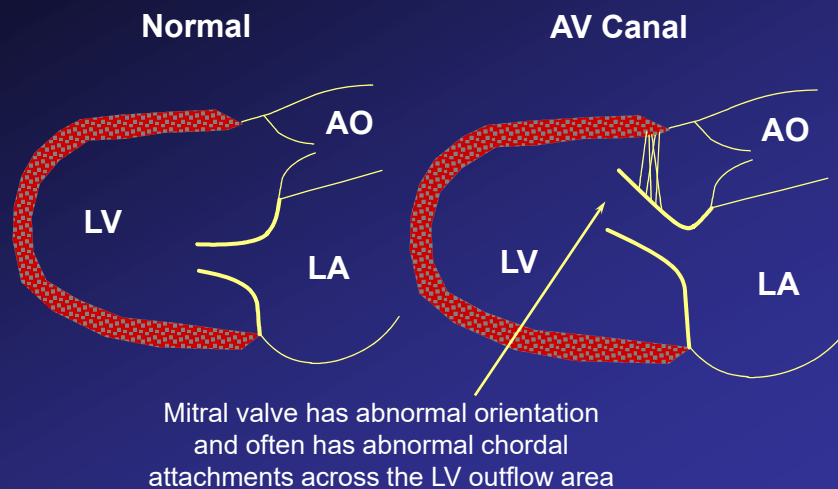
## Complete Atrioventricular Canal

### Associated Anomalies

- Patent ductus arteriosus
- Hypoplasia of one ventricle
- AV valve problems - regurgitation
- LVOT obstruction

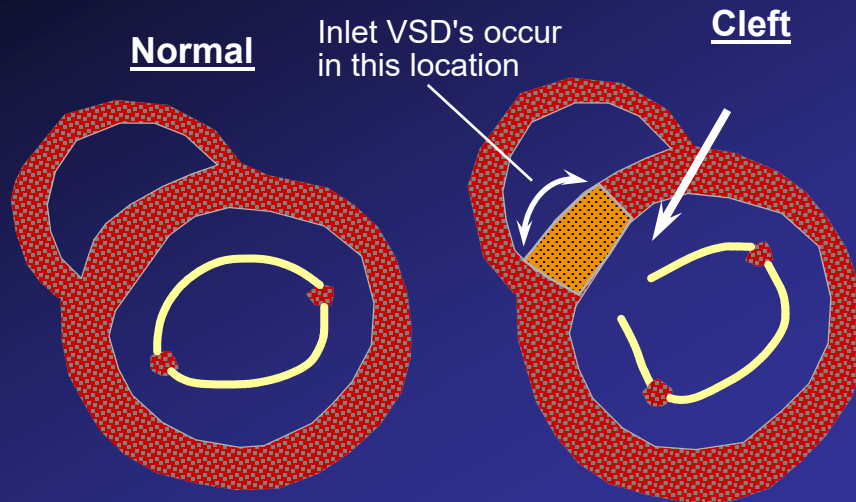
## Atrioventricular Canal

### Long Axis View



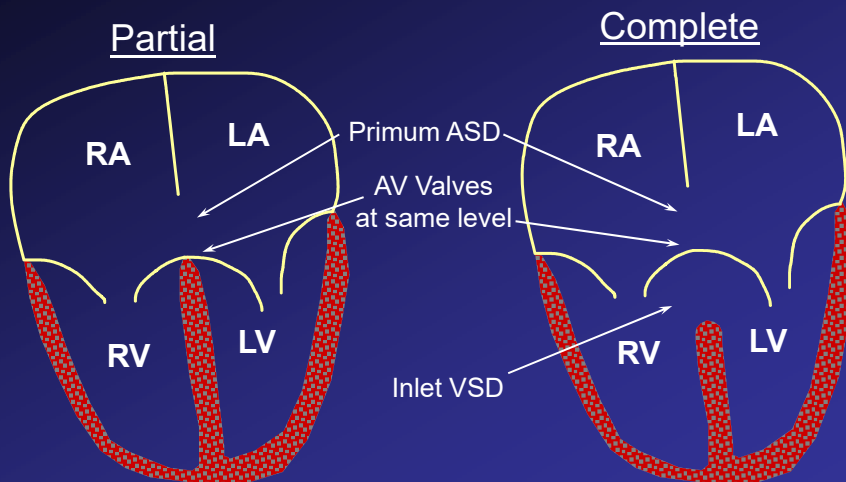
## Cleft Mitral Valve

Parasternal Short-Axis View



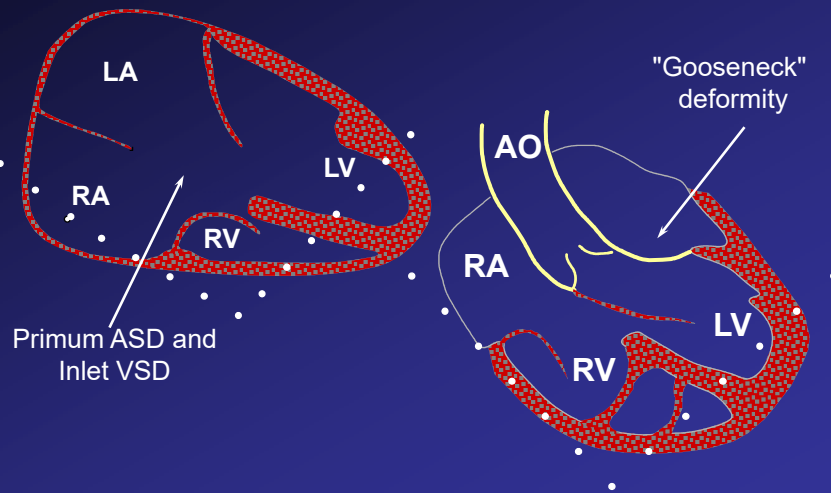
## Atrioventricular Canal

Apical 4-Chamber View



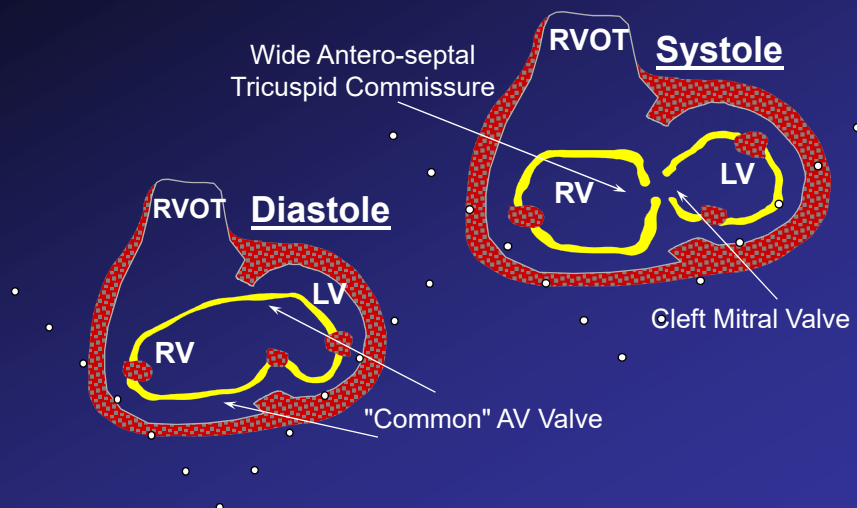
## Atrioventricular Canal Defects

### Subcostal Views

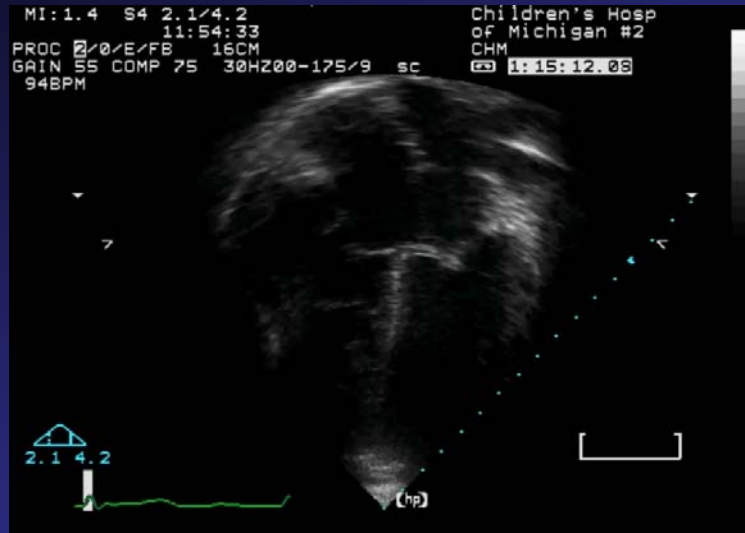


## Atrioventricular Canal Defects

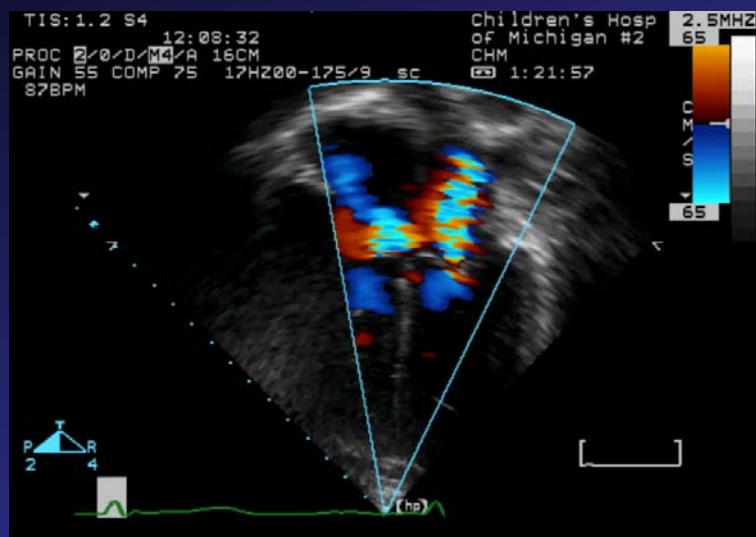
### Subcostal Short Axis Views



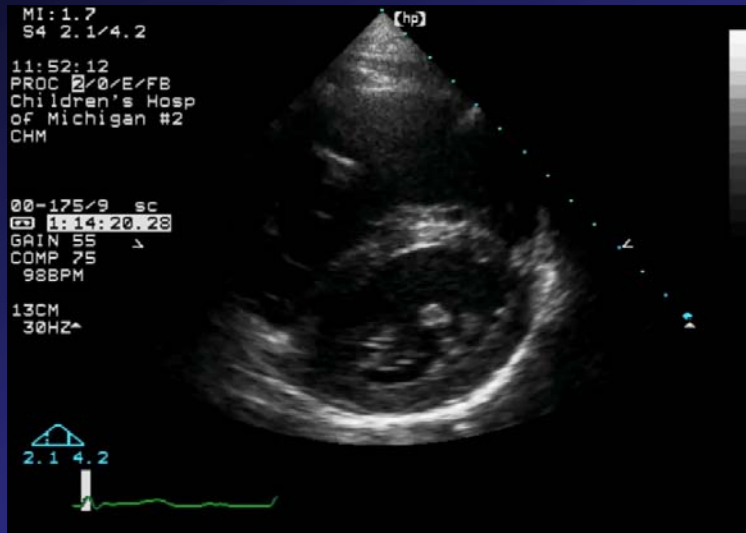
## Case 8 – Review



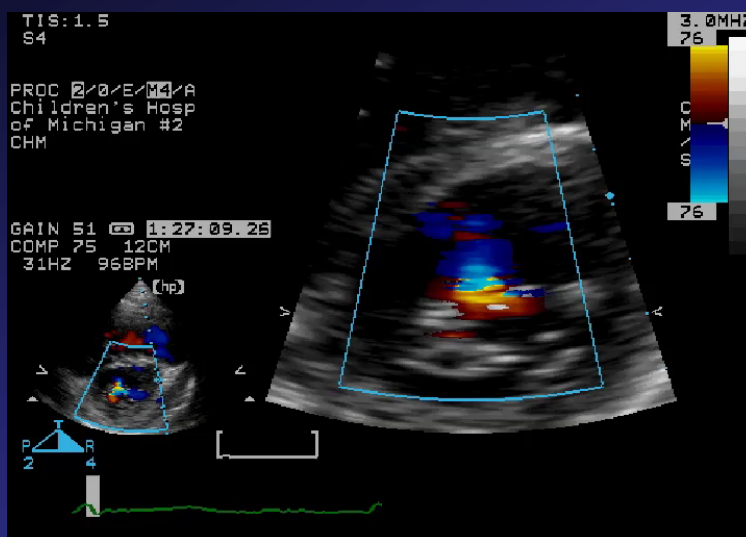
## Case 8 – Review



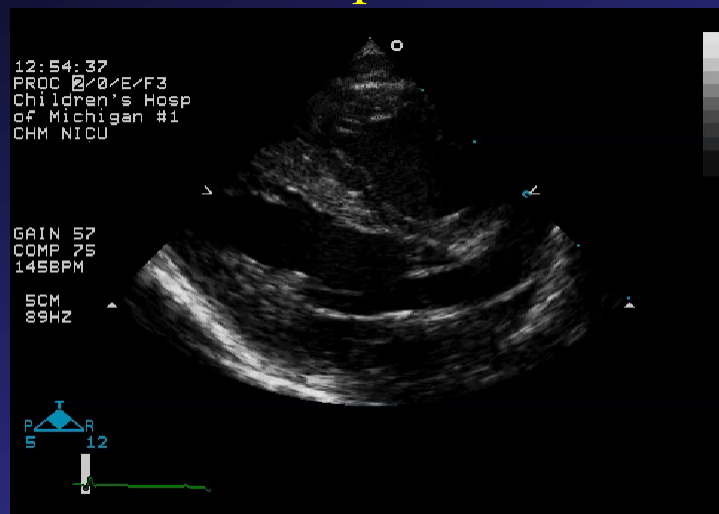
## Case 8 – Review



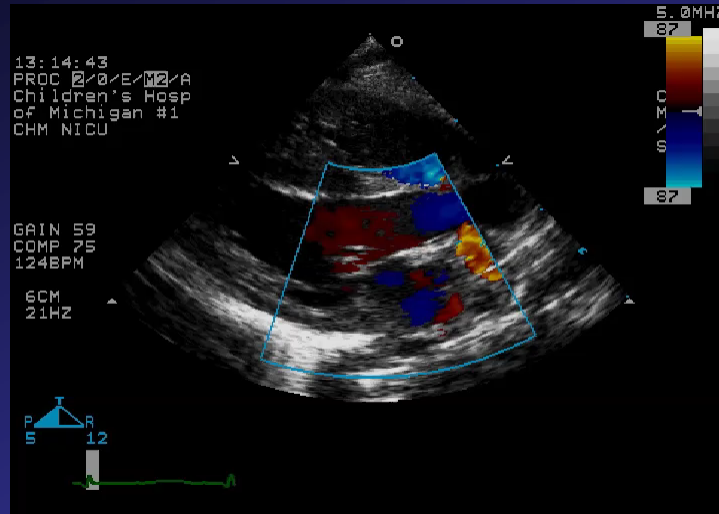
## Case 8 – Review



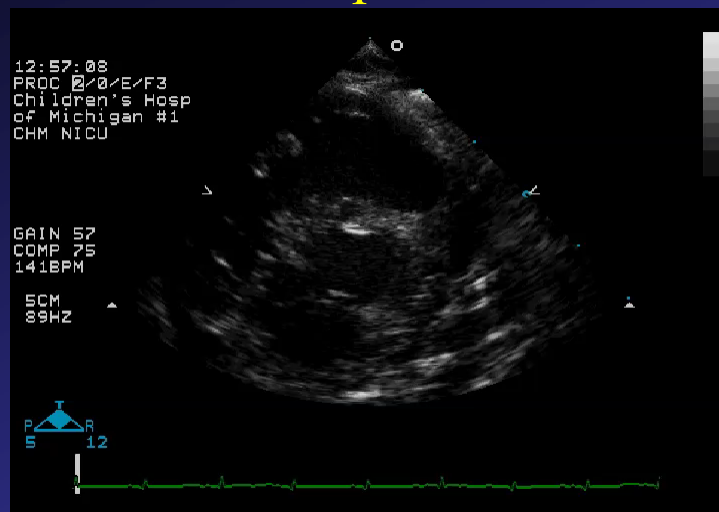
## Case 9 – Complete AV Canal



## Case 9 – Complete AV Canal

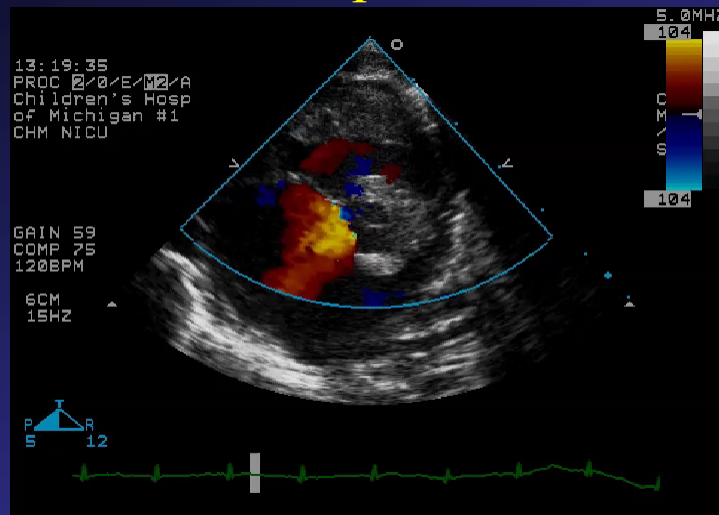


## Case 9 – Complete AV Canal





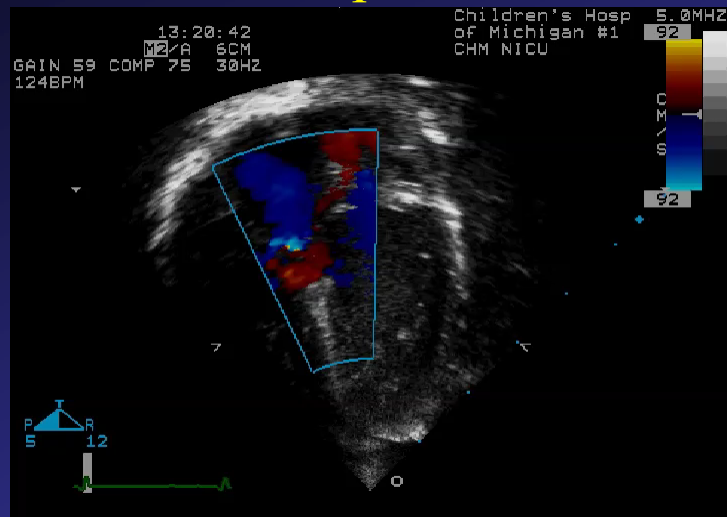
## Case 9 – Complete AV Canal



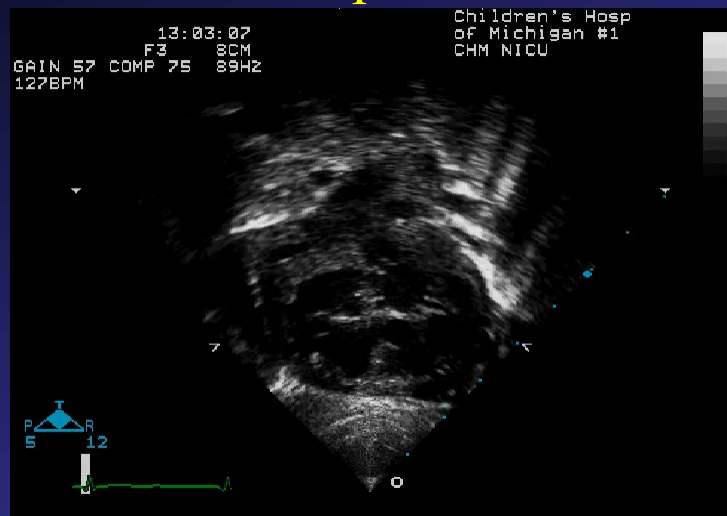
## Case 9 – Complete AV Canal



## Case 9 – Complete AV Canal



## Case 9 – Complete AV Canal



# Physiology

- 71

## AV Septal Defects

### Surgical Intervention

- Partial AVSD
  - Usually electively repaired at age 2-4 years
  - Complicating features (AVV regurgitation, LVOTO) may necessitate earlier intervention
- Complete AVSD
  - Usually repaired by 6 months of age (earlier in trisomy 21) to prevent pulmonary vascular obstructive disease

## AV Septal Defects

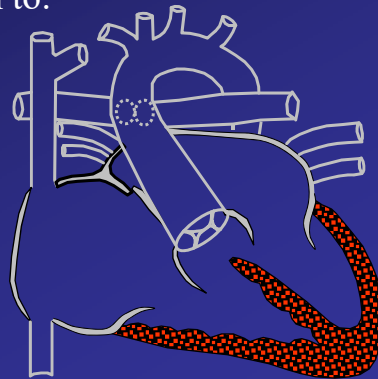
### Outcomes After Surgery

- Surgical outcomes
  - 3-4% operative mortality for complete AVSD
  - <1% operative mortality for partial AVSD
- Late reoperation in 10-15%
  - Left AV valve regurgitation
  - Left ventricular outflow tract obstruction
    - ♥ Hypoplasia of the outflow tract
    - ♥ Accessory AV valve tissue
    - ♥ Discrete subaortic membrane

## Truncus Arteriosus

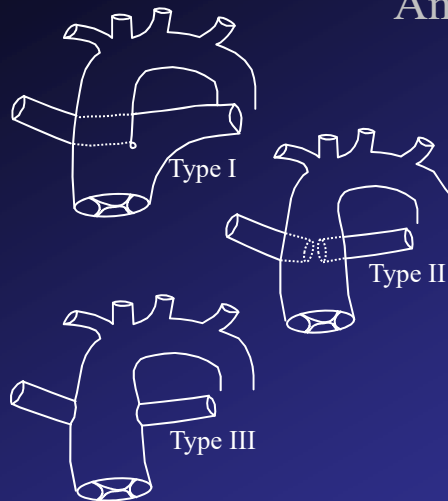
### Anatomy

- Characteristic anatomy characterized by:
  - Single arterial vessel that arises from the base of the heart and gives origin to:
    - ♥ Systemic arteries
    - ♥ Pulmonary arteries
    - ♥ Coronary arteries
  - Single semilunar valve



## Truncus Arteriosus

### Anatomy



### Associated Defects

- Abnormal coronaries (37-49%)
- Right aortic arch (30%)
- Abnormal truncal valve
- Absent pulmonary artery (16%)
- Interrupted aortic arch (15%)
- Left SVC (12%)
- Secundum ASD (9-20%)

## Truncus Arteriosus

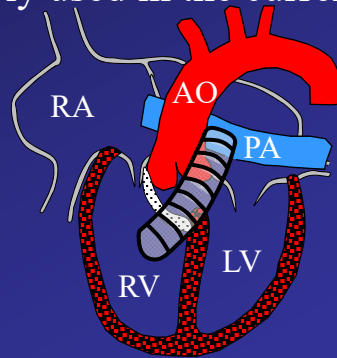
### Clinical Aspects

- Patients usually present due to the presence of a cardiac murmur
- Complete mixing of systemic and pulmonary venous blood results in cyanosis
- Excessive pulmonary blood flow causes sign and symptoms of congestive heart failure
- The cyanosis is generally mild

## Truncus Arteriosus

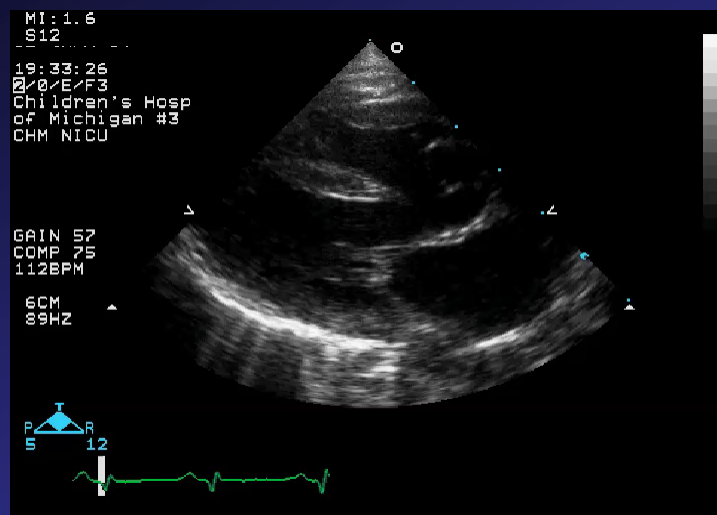
### Treatment

- Requires surgical repair in the first weeks of life
- Median sternotomy requiring bypass
- Palliative PA band rarely used in the current era
- Rastelli type repair
  - Close VSD to truncus
  - Disconnect PAs
  - RV to PA conduit



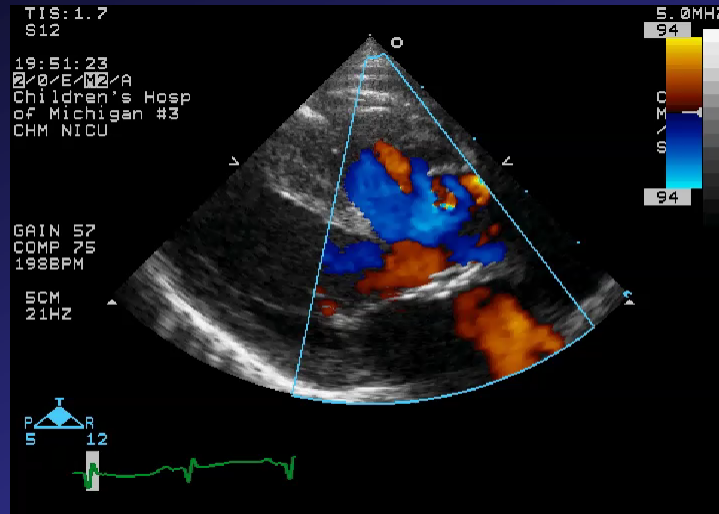
## Case 10

### Truncus Arteriosus



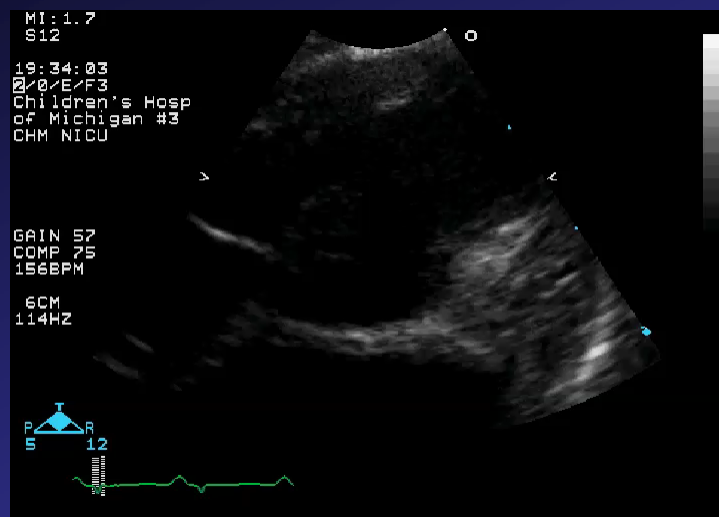
## Case 10

### Truncus Arteriosus



## Case 10

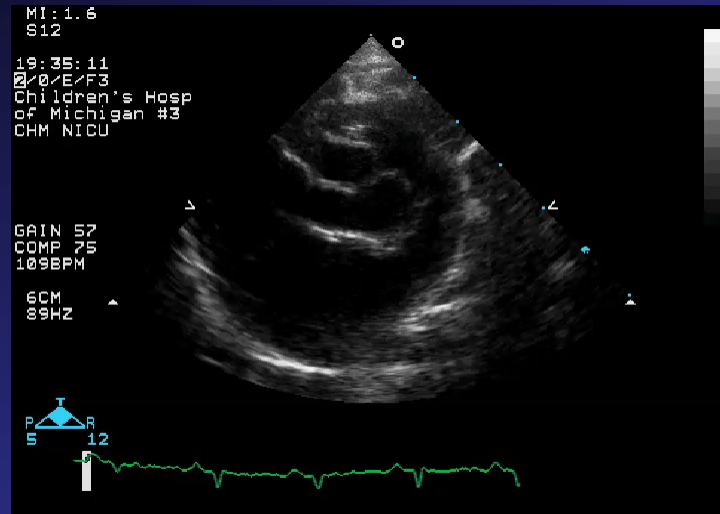
### Truncus Arteriosus





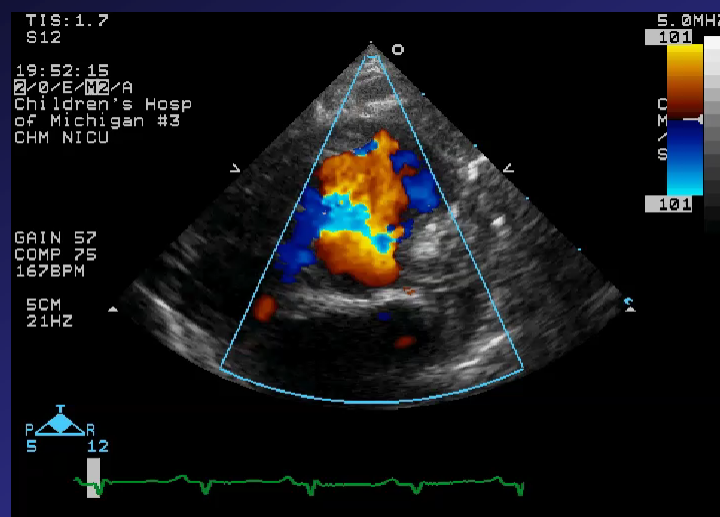
## Case 10

### Truncus Arteriosus



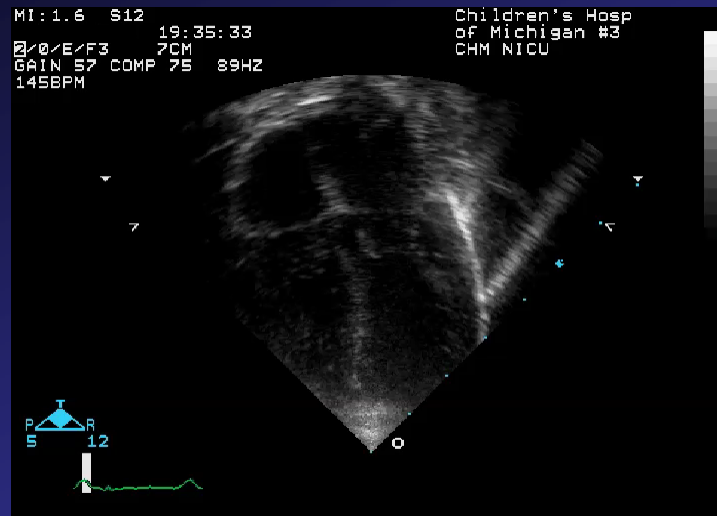
## Case 10

### Truncus Arteriosus



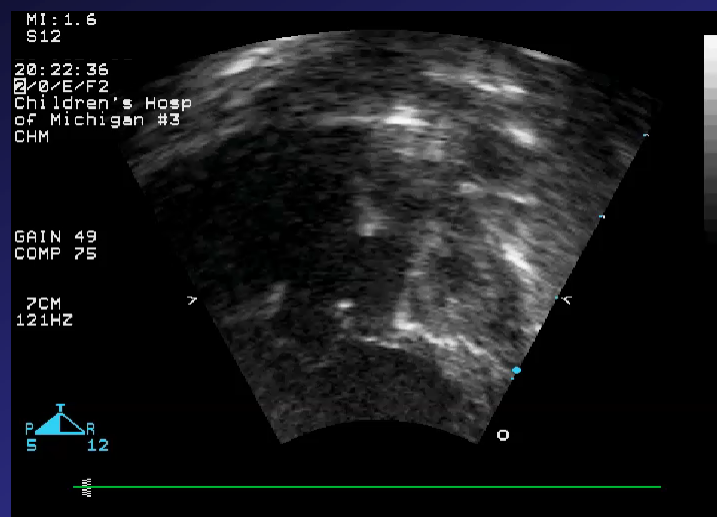
## Case 10

### Truncus Arteriosus



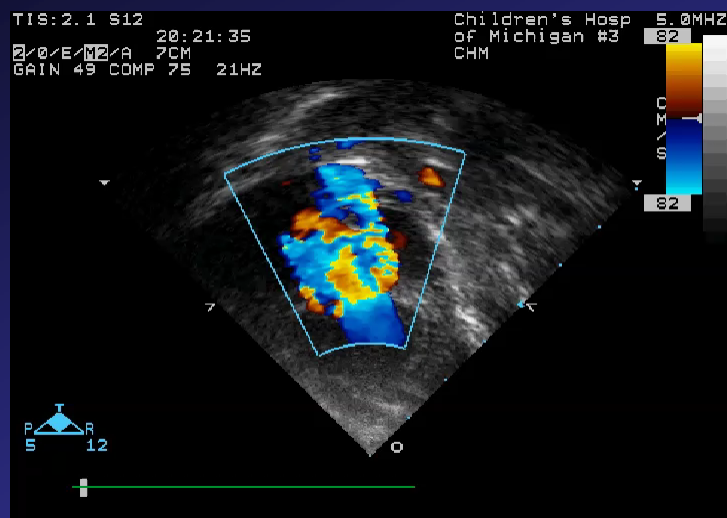
## Case 10

### Truncus Arteriosus



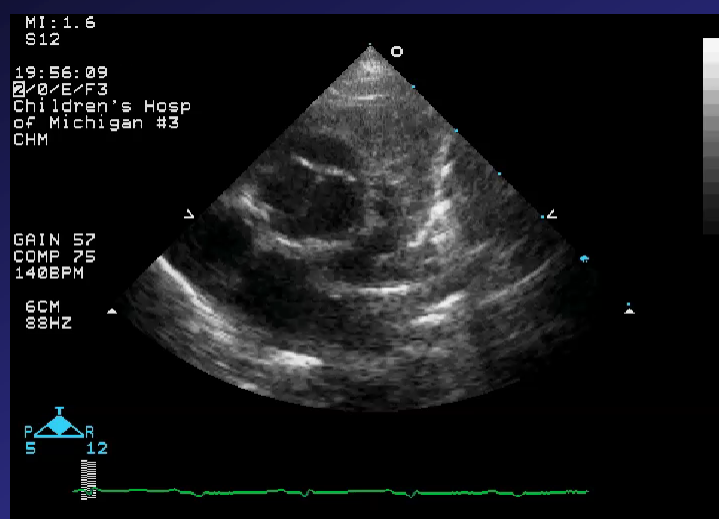
## Case 10

### Truncus Arteriosus



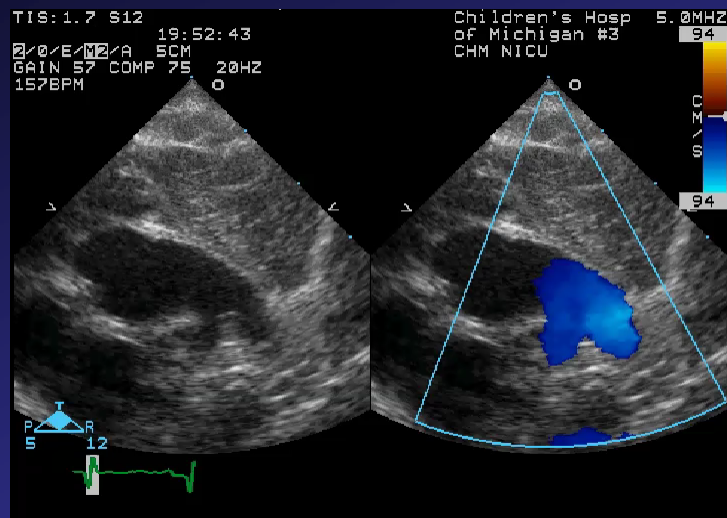
## Case 10

### Truncus Arteriosus



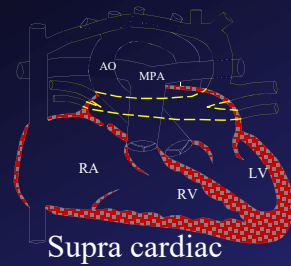
## Case 10

### Truncus Arteriosus

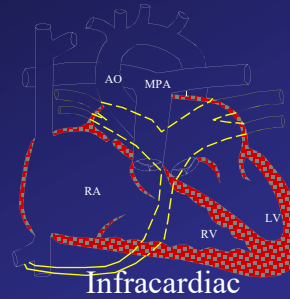


## Total Anomalous Pulmonary Venous Return

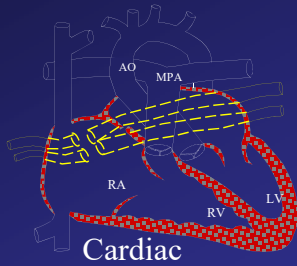
### Anatomic Types



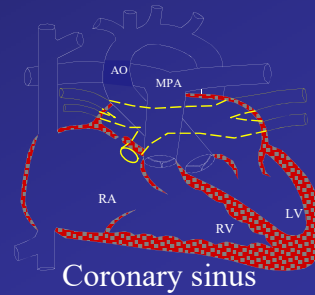
Supra cardiac



Infracardiac



Cardiac



Coronary sinus

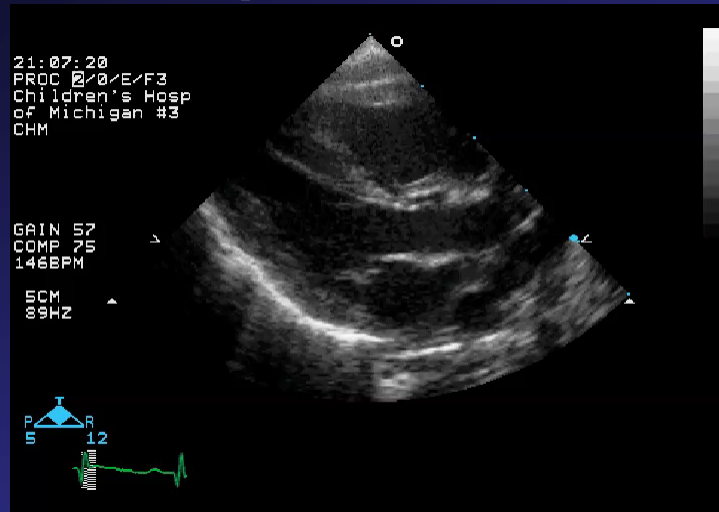
## Total Anomalous Pulmonary Venous Return

### Echo Clues

- Enlarged right heart
- Right to left atrial shunting
- Unusual “membranes” in left atrium
- Abnormal flow in systemic venous system
- Obstruction may occur at different levels
  - Most common - infracardiac
- May be remarkably asymptomatic (in absence of obstruction)

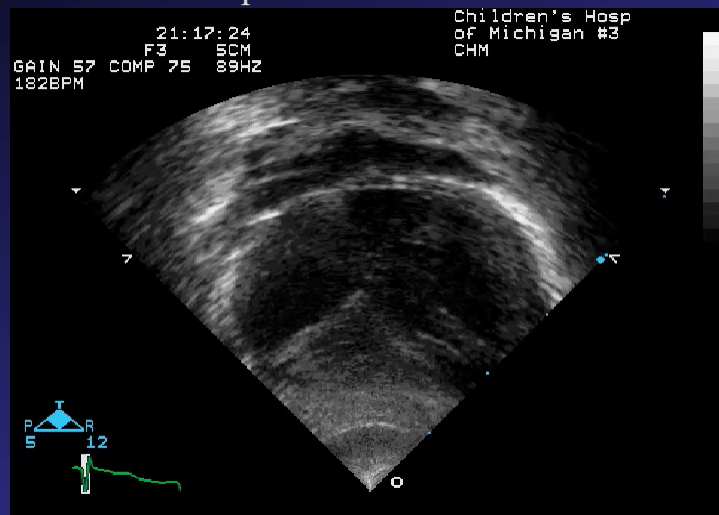
## Case 12

Supra cardiac TAPVR



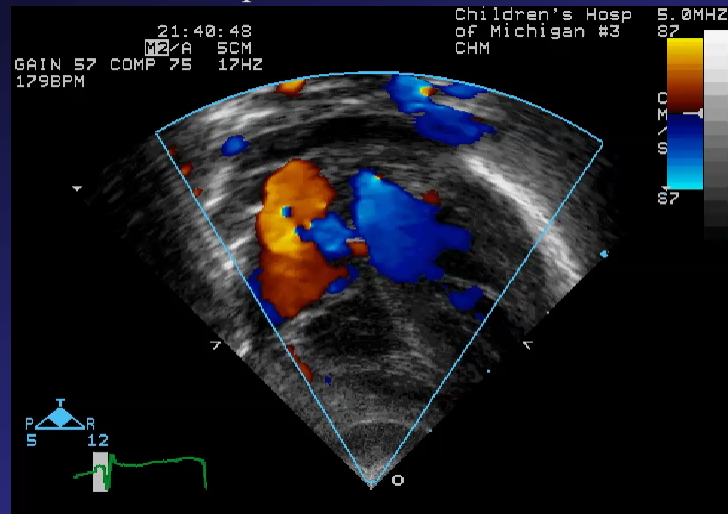
## Case 12

Supra cardiac TAPVR



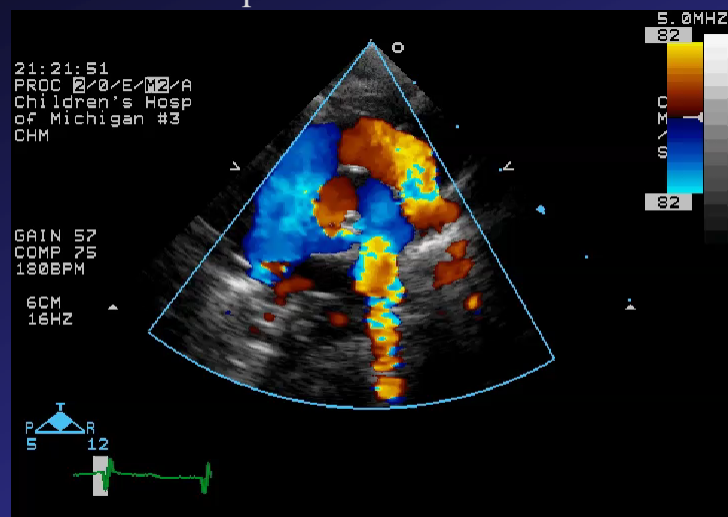
## Case 12

### Supra cardiac TAPVR



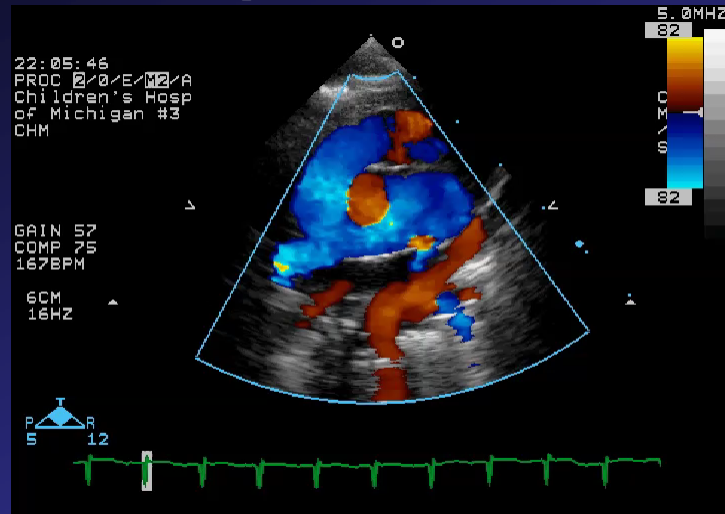
## Case 12

### Supra cardiac TAPVR



## Case 12

### Supra cardiac TAPVR





## Anomalous Left Coronary Artery

From the Pulmonary Artery - ALCAPA



- Rare congenital anomaly
- Usually presents at 2-3 m of age
- Results in severe LV ischemia
- Present as dilated CM, CHF
- ECG often diagnostic
- Patients survive w/ collateral flow
- Surgery done to re-implant vessel

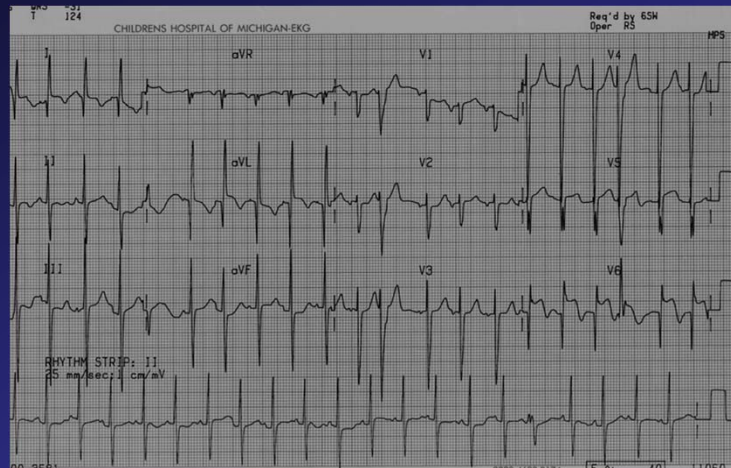
## Anomalous Left Coronary Artery

Echocardiographic Clues

- Left ventricular dysfunction (usually severe)
- Mitral insufficiency – due to LV dilation/dysfunction, papillary muscle infarction
- Endocardial fibroelastosis of LV and/or papillary muscles
- Failure to identify proximal LCA from aorta
- Unusual flow into main pulmonary artery

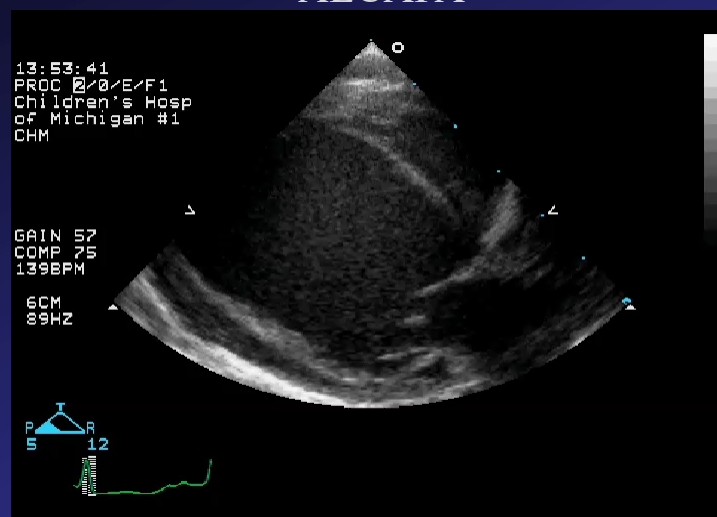
# Anomalous Left Coronary Artery

From the Pulmonary Artery - ALCAPA



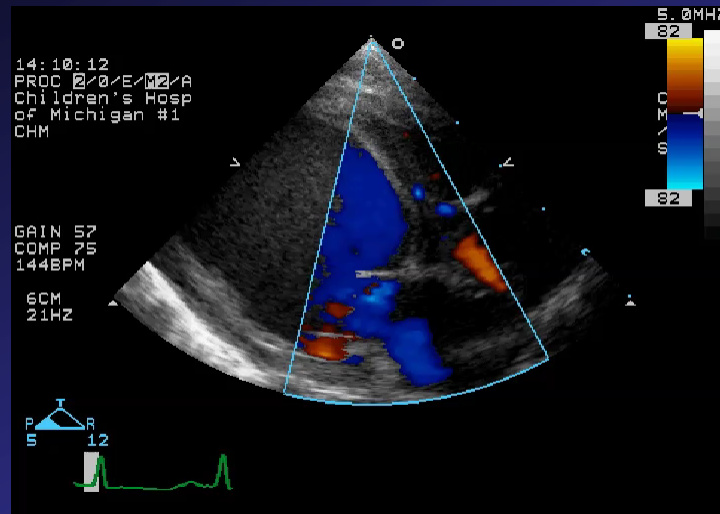
## Case 11

ALCAPA



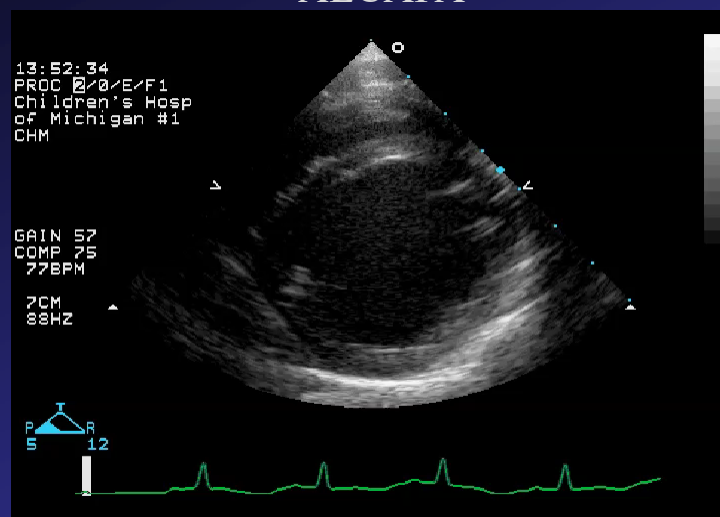
## Case 11

### ALCAPA



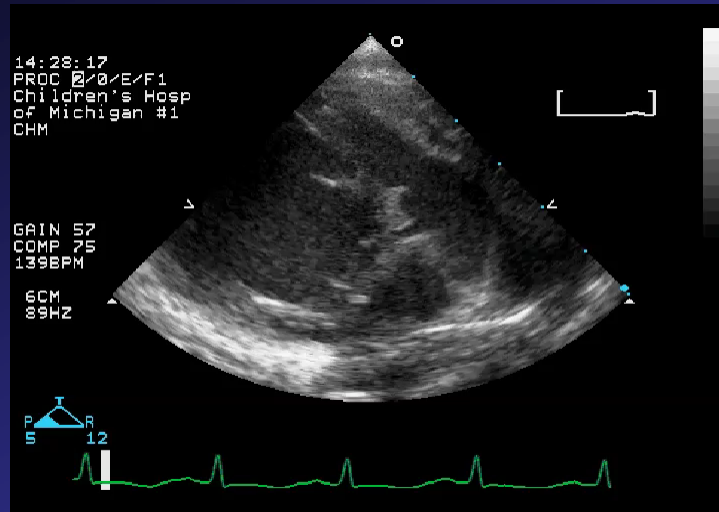
## Case 11

### ALCAPA



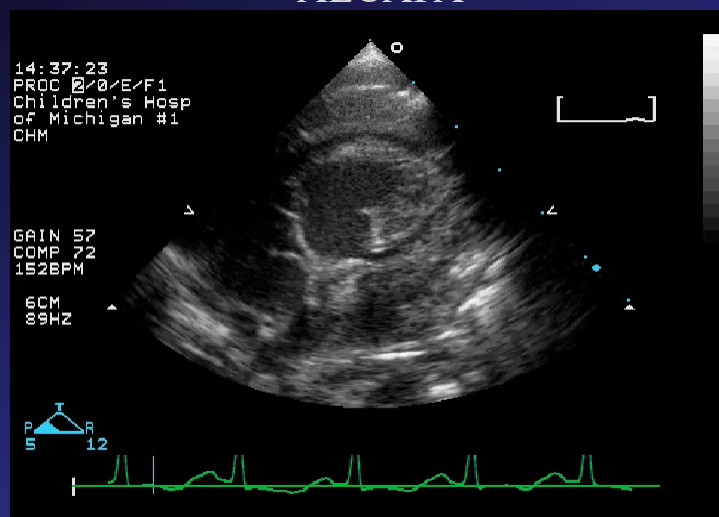
## Case 11

### ALCAPA



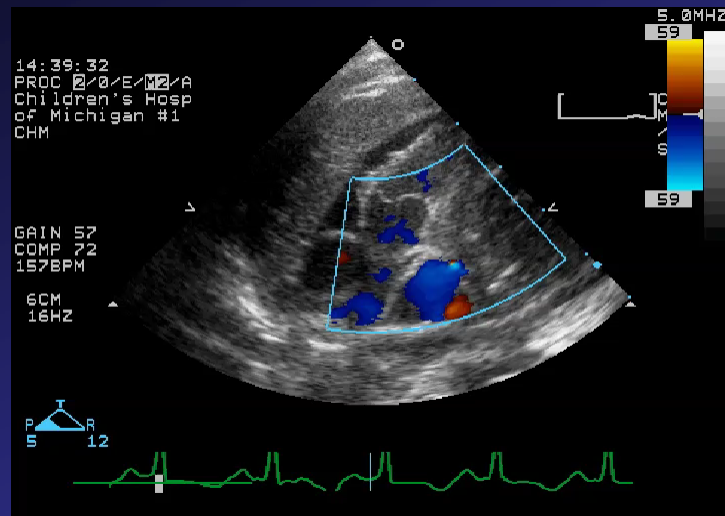
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**Good Luck On Your Exam**



**Questions?**

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